

## Supplementary Appendix

This appendix has been provided by the authors to give readers additional information about their work.

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## Supplementary Appendix

### *SF3B1* and Other Novel Cancer Genes in Chronic Lymphocytic Leukemia

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## MATERIALS AND METHODS

### **Human Samples**

Heparinized blood samples and skin biopsies were obtained from normal donors and patients enrolled on clinical research protocols that were approved by the Human Subjects Protection Committee at the Dana-Farber Cancer Institute (DFCI). In some cases, 2 ml of saliva was collected from study participants as a source of normal epithelial cell DNA. Peripheral blood mononuclear cells (PBMC) from normal donors and patients were isolated by Ficoll/Hyque density gradient centrifugation. CD19+ B cells from normal volunteers were isolated by immunomagnetic selection (Miltenyi Biotec, Auburn CA). Mononuclear cells were used fresh or cryopreserved with FBS 10% DMSO and stored in vapor-phase liquid nitrogen until the time of analysis. Primary skin fibroblast lines were generated from five mm diameter punch biopsies of skin that were provided to the Cell Culture Core lab of the Harvard Skin Disease Research Center, as previously described<sup>1</sup>. Second or third passage cultures were used for genomic DNA isolation.

### **Prognostic factor analysis**

Immunoglobulin heavy-chain variable (IGHV) homology (high risk unmutated was defined as greater than or equal to 98% homology to the closest germline match) and ZAP-70 expression (high risk positive defined as >20%) were determined as previously described<sup>2</sup>. Cytogenetics were evaluated by FISH for the most common CLL abnormalities (del(13q), trisomy 12, del(11q), del(17p), rearrangements of chromosome 14; all probes from Vysis, Des Plaines, IL) at the Brigham and Women's Hospital

Cytogenetics Laboratory, Boston MA<sup>3</sup>. Samples were scored positive for a chromosomal aberration based on consensus cytogenetic scoring<sup>4</sup>. Percent tumor cells harboring common CLL cytogenetic abnormalities, detected by FISH cytogenetics, are tabulated per sample in Table 9 Supplemental Appendix.

### **Whole-genome and -exome DNA sequencing**

Informed consent on DFCI IRB-approved protocols for whole genome sequencing of patients' samples was obtained prior to the initiation of sequencing studies. Genomic DNA was isolated from patient CD19<sup>+</sup>CD5<sup>+</sup> tumor cells and autologous skin fibroblasts (Wizard kit; Promega, Madison WI) per manufacturer's instructions. Alternatively, germline genomic DNA was extracted from autologous epithelial cells, obtained from saliva samples (DNA Genotek, Kanata, Ontario, Canada) or from autologous blood granulocytes, isolated following Ficoll/Hypaque density gradient centrifugation.

Whole genome shotgun (WG) and whole exome (WE) capture libraries were constructed as previously described<sup>5-7</sup>. For 51 (56%) of the 91 CLL samples included in the analysis, sequencing was performed on capture libraries generated from whole genome amplified (WGA) samples. For those samples, 100 ng inputs of samples were whole genome amplified with the Qiagen REPLI-g Midi Kit (Valencia, CA). No significant differences in mutation rate were observed between data originating from WGA and non-WGA samples (see Table 2 in the Supplemental Appendix). WGS libraries were sequenced on an average of 39 lanes of an Illumina GA-II sequencer, using 101 bp paired-end reads,

with the aim of reaching 30X genomic coverage of distinct molecules per sample<sup>6, 7</sup>. Exome sequencing libraries were sequenced on three lanes of the same instrument, using 76 bp paired-end reads.

Sequencing data subsequently was processed using the “Picard” pipeline, developed at the Broad Institute’s Sequencing Platform (Fennell T, unpublished; Cambridge, MA), which includes base-quality recalibration<sup>8</sup>, alignment to the NCBI Human Reference Genome Build hg18 using MAQ<sup>9</sup>, and aggregation of lane- and library-level data.

### **Identification of somatic tumor mutations and calculation of significance**

From the sequencing data, tumor-specific gene alterations were identified using a set of tools contained with the “Firehose” pipeline<sup>6, 7</sup>, developed at the Broad Institute. Somatic single nucleotide variations (SSNVs) were detected using muTect (Cibulskis et al, in preparation), while somatic small insertions and deletions were detected using the algorithm Indelocator (Sivachenko el al, in preparation). We applied the algorithm MutSig (Lawrence in preparation; <sup>10-12</sup>) to sequencing data from the 3 genomes and 88 exomes. Briefly, MutSig tabulates the number of mutations and the number of adequately covered bases for each gene (i.e. bases with  $\geq 14$  tumor and  $\geq 8$  normal reads). The counts are broken down by mutation context category (i.e. CpG transitions, other C:G transitions, any transversion, A:T transitions). For each gene, we calculate the probability of seeing the observed constellation of mutations or a more extreme one, given the background mutation rates calculated across the dataset (see Table 3 in the Supplemental Appendix for background mutation rate). This is done by convoluting a set of binomial distributions as

described previously, which results in a p and q value<sup>12</sup>. We observed that the 4 samples for which normal germline DNA was derived from blood granulocytes had a significantly lower detection of somatic mutations, suggesting contamination with tumor DNA. Reanalysis excluding these 4 samples had little affect on mutation rate (increased by only 5%: 0.71 mutations/Mb to 0.75 mutations/Mb) and yielded the same results of significantly mutated genes ( $q < 0.1$ ). All mutations in genes that were significantly mutated or within pathways related to these significantly mutated genes were confirmed by manual inspection of the sequencing data<sup>13</sup>. Furthermore, these mutations were also validated using an independent platform (Sequenom mass spectrometry-based genotyping). We saw no significant difference in non-synonymous mutation rate between IGHV-mutated and –unmutated patients (despite 82% power to detect differences of 0.6 standard deviations; one-sided 0.05 level test) or between different clinical stages. Our ability to detect mutations of low allele fraction depends on several factors, including the purity and ploidy of the sample, and the copy number at the locus in question. Graphical representation of the distribution of allelic fraction among the total number of 2348 mutations detected is depicted in Figure 7 in the Supplemental Appendix. To estimate our rate of false-positive mutation calls, we randomly chose a subset of our putative somatic point mutations and indels to be subjected to orthogonal validation by multiplexed Sequenom mass spectrometry assays. Because of the limited sensitivity of this assay at low allele fractions, we restricted our analysis to mutations that were present in the tumor at an allele fraction of at least one-third. We designed Sequenom assays for 71 randomly selected mutations, and of these, 66 were successfully validated as somatic. The other 5 were deemed to be reference. This yields an estimated specificity of 93%.

### **Statistical analysis of mutation rate in association with clinical variables**

Clinical data were available from 91 CLL samples comprising the genome/exome sequenced discovery set, and from 101 CLL samples used for extension and validation. We tested the association between patient characteristics and clinical variables such as time to first treatment (TTFT), and mutation rate or presence or absence of driver mutations. P-values were calculated using the Wilcoxon rank sum test for quantitatively measured variables across two groups, the Fisher Exact test for categorical variables, the Kruskal-Wallis test for quantitatively measured variables across three groups and for ordered categorical data, and the log rank test for comparing Kaplan-Meier estimated censored time to event variables. Time to first therapy was defined as the elapsed time between initial diagnosis and first treatment for CLL. Patients who remained untreated for their disease at the most recent follow-up were censored at that time. All statistical tests were performed using SAS software version 9.2 and R version 2.8.0.

Univariate analysis was performed using Cox proportional hazards regression for the 19 variables potentially predictive of TTFT including (IGHV mutated vs. unmutated vs. unknown, ZAP-70 negative vs. positive vs. unknown, Rai stage at sampling 0/1 vs 2/3/4 vs unknown, age ( $\geq 55$  yrs. vs.  $< 55$  yrs), sex, presence of del(17p), del(11q), trisomy(12), homozygous del(13q), heterozygous del(13q), presence of mutations in *ATM*, *NOTCH1*, *SF3B1*, *TP53*, *DDX3X*, *ZMYM3*, *MAPK1*, *FBWX7*, *MYD88*). A stepwise Cox proportional hazards regression model of TTFT was performed for the 91 discovery samples, using the 19 variables listed above. The same final model was obtained with a forward selection

procedure. Step-up models using the -2 log likelihood statistic to assess goodness of fit using the appropriate degrees of freedoms were also explored. Cox modeling results are reported as hazard ratios along with the 95% confidence intervals.

### **Detection of altered RNA splicing**

Total RNA was extracted from normal B and CLL-B cells (TRIZOL; Invitrogen, Carlsbad CA). 2 $\mu$ g total RNA from each sample was treated with DNase I (2 units/sample; New England BioLabs, Ipswich MA) at 37°C for 20 minutes to remove contaminating genomic DNA, followed by heat-inactivation of DNase I at 75°C for 15 minutes, and then used as template to synthesize cDNA by reverse transcription (SuperScript® III First-Strand kit; Invitrogen, Carlsbad CA). We designed in parallel quantitative Taqman assays primers to detect spliced transcripts across consecutive exons, and unspliced transcripts in which one primer was localized within the retained intron. Details of primer design the splicing assays for RIOK3, and BRD2 are noted in Table 10 of Supplemental Appendix. All assays were run in triplicate using the 7500 Fast System (Applied Biosystems, Carlsbad CA), and all values were normalized to GAPDH gene expression. Relative splicing activity was measured by calculating the ratio of unspliced to spliced forms of each target gene. For some experiments, splicing was measured following treatment of 293 cells or normal B cells or CLL cells with the SF3b-complex targeting drug E7107 at 1  $\mu$ M (gift of Robin Reed, HMS).

## SUPPLEMENTAL FIGURES AND LEGENDS

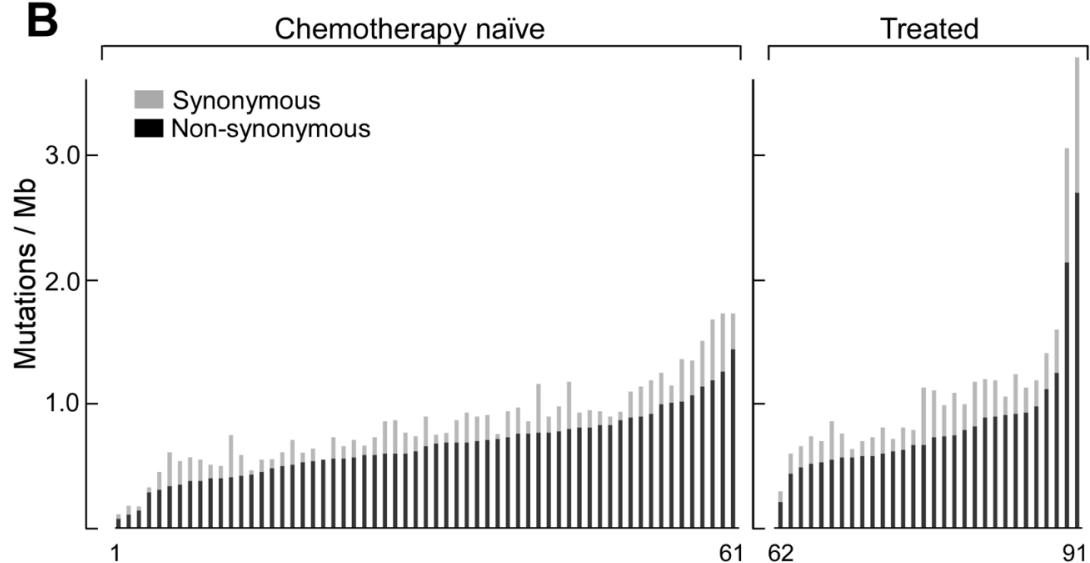
**Supplemental Figure 1. Mutation rate is unrelated to treatment status in CLL patients.** **A.** Clinical summary of the 91 patients sequenced. **B.** Mutation rate is similar between 61 chemotherapy-naïve and 30 chemo-treated CLL samples.

**A**

Clinical characteristics at time of sample (n = 91)

Median age (yrs)	54 (34,78)	Treatment status	<i>IGHV mutation status</i>
Sex	35F,56M	Chemo naive ....	Unmutated ..... 40
		Treated ..... 30	Mutated ..... 38
			Unknown ..... 13
Rai classification		ZAP-70	<i>Detected cytogenetic abnormality by FISH (%)</i>
0 .....	29	Negative .....	Del (13q), monoallelic 53(58)
1 .....	43	Positive .....	Del (13q), biallelic 12(13)
2 - 4 .....	19	Unknown .....	Trisomy 12 ..... 13(14)
			Del (11q) ..... 22(24)
			Del (17p) ..... 17(19)

**B**



**Supplemental Figure 2.** Mutations in *SF3B1*, *FBXW7*, *DDX3X*, *MAPK1*, *NOTCH1* and *ZMYM3* occur in evolutionarily conserved regions. For *SF3B1*, of the 14 novel mutations discovered in 91 CLL samples, all were localized to conserved regions of genes. Where available, alignments of gene sequences around each mutation are shown for human, mouse, zebrafish, *C.elegans* and *S.pombe* genes (USCS Genomic Bioinformatics: <http://genome.ucsc.edu>). A similar analysis was performed in the other significantly mutated genes.

			Repeat 4 (608-639)	R625L N626H	Repeat 5 (640-674)		
Human	569	PYVHKILVVIEPLLIDEDYYARVEGREIISNLAKAAGLATMISTMRPDIDNMDEYVRNTTARAFAVVASALGIPSLLPFL		••		648	
Mouse	569	PYVHKILVVIEPLLIDEDYYARVEGREIISNLAKAAGLATMISTMRPDIDNMDEYVRNTTARAFAVVASALGIPSLLPFL				648	
Zebrafish	580	PYVHKILVVIEPLLIDEDYYARVEGREIISNLAKAAGLATMISTMRPDIDNMDEYVRNTTARAFAVVASALGIPSLLPFL				659	
<i>C.elegans</i>	587	PYVHKILVVIEPLLIDEDYYARVEGREIISNLAKAAGLATMISTMRPDIDNVDEYVRNTTARAFAVVASALGIPSLAPPFL				666	
<i>S. pombe</i>	453	PTTHKILVVIEPLLIDEDYYARAEGREIISNLAKASGLAHMIATMRPDIDHVDEYVRNTTARAFSVVASALGVPALLPFL				532	
			Repeat 6 (675-723)	K700E	Repeat 7 (724-765)		
Human	649	KAVCKSKKSWQARHTGKIVQQIAILMGCALPLHRLRSILVEIIIEHGLVDEQQKVRTISALAALEAATPYGIESFDSVL		•	G740E K741N K742D	728	
Mouse	649	KAVCKSKKSWQARHTGKIVQQIAILMGCALPLHRLRSILVEIIIEHGLVDEQQKVRTISALAALEAATPYGIESFDSVL				728	
Zebrafish	660	KAVCKSKKSWQARHTGKIVQQIAILMGCALPLHRLRSILVEIIIEHGLVDEQQKVRTISALAALEAATPYGIESFDSVL				739	
<i>C.elegans</i>	667	KAVCKSKKSWQARHTGKIVQQMAILMGCALPLHKLALDIVEQLGLDEQQKVRTITALCLAALEAASSPYGIEAFDSVL				746	
<i>S. pombe</i>	533	KAVCRSKKSWQARHTGVRIIQQIAALLGCSILPHLKNLVDCIGHGLEDEQQKVRTIMTALSALAALEAATPYGIEAFDSVL				612	
			Repeat 8 (724-765)		Repeat 9 (806-844)	Repeat 10 (845-887)	
Human	729	KPLWKIGRQRKGKLAALFLKAIGYLIPILMDAEYANYYTREVMLLIREFQSPDEEMKKIVLKVKQCCGTGVEANYIKT	•••		EILPPFFKHFQHMRMALDRRNRYQLVDTTVELANKVGAAEIISRIVDDLKEAEQYRKVMVMETIEKIMGNLGAADIDHKL	808	
Mouse	729	KPLWKIGRQRKGKLAALFLKAIGYLIPILMDAEYANYYTREVMLLIREFQSPDEEMKKIVLKVKQCCGTGVEANYIKT				808	
Zebrafish	740	KPLWKIGRQRKGKLAALFLKAIGYLIPILMDAEYANYYTREVMLLIREFQSPDEEMKKIVLKVKQCCATDGVEANYIKT				819	
<i>C.elegans</i>	747	KPLWKIGRMHRKGKLAALFLKAIGYLIPILMDAEYASYYTREVMLLIREFASPDEEMKKIVLKVKQCCATDGVEASYIRD				826	
<i>S. pombe</i>	613	KPLWSGVQRHRGKSLAFLKATGFIPLMEPEYASHFTTRRIMKILLREFNSPDEEMKKIVLKVSQCASTDGVTPPEYLRT				692	
			Repeat 11 (886-923)	Q903R			
Human	809	EILPPFFKHFQHMRMALDRRNRYQLVDTTVELANKVGAAEIISRIVDDLKEAEQYRKVMVMETIEKIMGNLGAADIDHKL		•		EEQLIDGILYAFQEQTTEDSVMMLNGFGTVVNLGKRVKPYLPQICGTVLWRLNNKSAKVRQQAADLISRTAVVMKTCQEE	888
Mouse	809	EILPPFFKHFQHMRMALDRRNRYQLVDTTVELANKVGAAEIISRIVDDLKEAEQYRKVMVMETIEKIMGNLGAADIDHKL				EEQLIDGILYAFQEQTTEDSVMMLNGFGTVVNLGKRVKPYLPQICGTVLWRLNNKSAKVRQQAADLISRTAVVMKTCQEE	888
Zebrafish	820	EILPPFFKHFQHMRMALDRRNRYQLVDTTVELANKVGAAEIISRIVDDLKEAEQYRKVMVMETIEKIMGNLGAADIDHKL				EEQLIDGILYAFQEQTTEDSVMMLNGFGTVVNLGKRVKPYLPQICGTVLWRLNNKSAKVRQQAADLISRTAVVMKTCQEE	899
<i>C.elegans</i>	827	EVLESFFKAFWNORMADMRRNYRQLVDTTVEIAQKVGCVERMIARIVDDLKDENEQYRKVMVMETIEENIVALQGATDIDARL				EEQLIDGILYAFQEQTTEDSVMMLNGFGTVVNLGKRVKPYLPQICGTVLWRLNNKSAKVRQQAADLIRIAFPVMHCEEE	906
<i>S. pombe</i>	693	DVLPEFFFHCFWSRRMASDRRSYKQVVTETTVLAQQVGSRQIVERVVNNFKDESEPYRKMTAETVDKVGSLGVSEIDERL					772

# FBXW7

p.F280L

Human	234	LQEWLKMFQSWSGPEKLLALDELIDSCEPTQVKHMMQVIEPQFQRDFISLLPKELALYVLSFLEPKDQQAAQTCRYWRI	313
Mouse	237	LQEWLKMFQSWSGPEKLLALDELIDSCEPTQVKHMMQVIEPQFQRDFISLLPKELALYVLSFLEPKDQQAAQTCRYWRI	316
Zebrafish	132	LQEWLRTFQSWSGPEKLLALDELIDSCEPTQVKHMMQVIEPQFQRDFISLLPRELALHVLSFLEPKDQQAAQTCRYWRI	211

Human	314	LAEDNLLWREKCKEEGIDEPLHIKRRKVIKPGFIHSPWKSAYIRQHRIDTNWRGELKSPKVLKGHDHVITCLQFCGNR	393
Mouse	317	LAEDNLLWREKCKEEGIDEPLHIKRRKVIKPGFIHSPWKSAYIRQHRIDTNWRGELKSPKVLKGHDHVITCLQFCGNR	396
Zebrafish	212	LAEDNLLWKEKCKEEGIDEPLHIKRRKVIKPGFTHSPWKSAYIRQHRIDTNWRGDLKSPKVLKGHDHVITCLQFCGNR	291

p.R465H

Human	394	IVSGSDDNTLKVWSAVTGKCLRTLTVGHTGGVWSSQMRDNIIISGSTDRTLKVWNAAETGEICIHTLYGHTSTVRCMHLHEKR	473
Mouse	397	IVSGSDDNTLKVWSAVTGKCLRTLTVGHTGGVWSSQMRDNIIISGSTDRTLKVWNAAETGEICIHTLYGHTSTVRCMHLHEKR	476
Zebrafish	292	IVSGSDDNTLKVWSAVTGKCLRTLTVGHTGGVWSSQMRDNIIISGSTDRTLKVWNAAETGEICIHTLYGHTSTVRCMHLHEKR	371

p.R505C

Human	474	VVSGSRDATALRVWDIETGQCLHVLMGHVAAVRCVQYDGRRVSGAYDFMVKVWDPEETCLHTLQGHTNRVYSLQFDGIH	553
Mouse	477	VVSGSRDATALRVWDIETGQCLHVLMGHVAAVRCVQYDGRRVSGAYDFMVKVWDPEETCLHTLQGHTNRVYSLQFDGIH	556
Zebrafish	372	VVSGSRDATALRVWDIETGQCLHVLMGHVAAVRCVQYDGRRVSGAYDFMVKVWDPEETCLHTLQGHTNRVYSLQFDGIH	451

p.G597E

Human	554	VVSGSLDTSIRVWDVETGNCIHTLTGHQSLTSGMELKDNILVSGNADSTVKIWIDIKTGQCLQLQGPNKHQSAVTCLQFN	633
Mouse	557	VVSGSLDTSIRVWDVETGNCIHTLTGHQSLTSGMELKDNILVSGNADSTVKIWIDIKTGQCLQLQGPNKHQSAVTCLQFN	636
Zebrafish	452	VVSGSLDTSIRVWDVETGNCIHTLTGHQSLTSGMELKDNILVSGNADSTVKIWIDIKTGQCLQLQGPNKHQSAVTCLQFN	531

# DDX3X

p.S24\*

Human	1	MSHVAVENALGLDQQFAGLDLNSSDNQSGGSTASKGRYIPPHLRNREATKGFYDKDSSGWSSSKDKDAYSSFGSRDSRG	80
Mouse	1	MSHVAVENALGLDQQFAGLDLNSSDNQSGGSTASKGRYIPPHLRNREATKGFYDKDSSGWSSSKDKDAYSSFGSRDSRG	80

Human	81	KSSFFSDRGSGSRGRFDRGRSDYDGIGGRGDRSGFGKFERGGNSRWCDKSDEDDWSKPLPPSERLEQEELFSGGNTGINF	160
Mouse	81	KSSFFGDRGSGSRGRFDRGRGDYDGIGGRGDRSGFGKFERGGNSRWCDKSDEDDWSKPLPPSERLEQEELFSGGNTGINF	160

Human	161	EKYDDIPIVEATGNNCPHIESFSDVEMGEIIMGNIELTRYTRPTPVQKHAIPPIKEKRLDMACAQTGSGKTAFLPILS	240
Mouse	161	EKYDDIPIVEATGNNCPHIESFSDVEMGEIIMGNIELTRYTRPTPVQKHAIPPIKEKRLDMACAQTGSGKTAFLPILS	240

Human	241	QIYADGPGEALRAMKENGRYGRRKQYPISLVLAPTRRELAVQIYEEARKFSYRSRVRPCVYGGAEIGQQIRDLERGCHLL	320
Mouse	241	QIYADGPGEALRAMKENGRYGRRKQYPISLVLAPTRRELAVQIYEEARKFSYRSRVRPCVYGGAEIGQQIRDLERGCHLL	320

p.K342\_splice

Human	321	VATPGRVLVDMMERGKIGLDFCKYLVLDEADRLMDMGFEPQIRRIVEQDTMPPKGVRHTMMFSATFPKEIQMLARDFLDEY	400
Mouse	321	VATPGRVLVDMMERGKIGLDFCKYLVLDEADRLMDMGFEPQIRRIVEQDTMPPKGVRHTMMFSATFPKEIQMLARDFLDEY	400

p.S410fs

Human	401	IFLAVGRVGSTSENITQKVVVVEESDKRSFLDLLNATGKDSLTLVVFETKKGADSLEDFLYHEGYACTSIHGDRSQRDR	480
Mouse	401	IFLAVGRVGSTSENITQKVVVVEEIDKRSFLDLLNATGKDSLTLVVFETKKGADSLEDFLYHEGYACTSIHGDRSQRDR	480

## MAPK1

pD162N

Human	152	PSNLLNNTCDLKICDFGLARVADPDDHHTGFLTEYVATRWYRAPEIMLNSKGYTKSIDIWSVGCILAEMLSNRPPIPPGK	231
Mouse	150	PSNLLNNTCDLKICDFGLARVADPDDHHTGFLTEYVATRWYRAPEIMLNSKGYTKSIDIWSVGCILAEMLSNRPPIPPGK	229
Zebrafish	161	PSNLLNNTCDLKICDFGLARVADPDDHHTGFLTEYVATRWYRAPEIMLNSKGYTKSIDIWSVGCILAEMLSNRPPIPPGK	240
		p.D291G	
Human	232	HYLDQLNHILGILGSPSQEDLNCIINLKARNYLLSLPHKNKVPWNRLFPNADSKALDLLDKMLTFNPBKRIEVEQALAHF	311
Mouse	230	HYLDQLNHILGILGSPSQEDLNCIINLKARNYLLSLPHKNKVPWNRLFPNADSKALDLLDKMLTFNPBKRIEVEQALAHF	309
Zebrafish	241	HYLDQLNHILGILGSPSQEDLNCIINIKARNYLLSLPLRSKVPWNRLFPNADPKALDLLDKMLTFNPBKRIEVEEALAHF	320
		p.Y316F	
Human	312	YLEQYYDPSDEPIAEAPFKFDMELDDLPEKLKELIFEETARFQPGYRS	360
Mouse	310	YLEQYYDPSDEPIAEAPFKFDMELDDLPEKLKELIFEETARFQPGYRS	358
Zebrafish	321	YLEQYYDPTDEPVAEAPFKFDMELDDLPEKLKELIFEETARFQPGYRP	369

## Notch1

p2514fs

Human	2503	QVPEHPFLTPSPESPDPDWSSSSPHSNVSDWSEGVSPPSTMOSQIARIPEAFK	2555
Mouse	2479	QVPEHPFLTPSPESPDPDWSSSSPHSNISDWSEGISSPPTTMPSQITHIPEAFK	2531
Zebrafish	2387	QVPDHFLTPSAGSPDQWSSSPHSNLSDWSEGISSPPTSM--QMNHIPEAFK	2437
Drosophila	2538	QVPDHFLTPSPEPDQWSSSPHSNVSDWSEGISSPPTSMOSQMGHIPEAFK	2590

## ZMYM3

53fs

Human	1	MDPSDFPSPFDPLTLPEKPLAGDLPVDMEFGEDLLESQTAPTRGWAPPGPSPSGALDLLDTFAGLEKDPG-VLDGATEL	79
Mouse	1	MDPSDFPSPFDPLTLPEKPLAGDLPVDMEFGEDLLESQTAPSRGWAPPGPSPSGALDLLDTFAGLEKDPGGVLDGATEL	80
Zebrafish	1	MSTEDFI-----GGTAESGKMDTTE-----	21
		Q399*	
Human	395	RPIPQSGDPADATRCSICQKTGEVL-HEVSNGSVVHRLCSDSCFSKFRANKGLKTNCCDQCGAYIYTKTGSPGP-ELLFH	472
Mouse	397	RPIPQSGDPADATRCSICQKTGEVL-HEVSNGSVVHRLCSDSCFSKFRANKGLKTNCCDQCGAYIYARPGGLGP-ELLFH	474
Zebrafish	265	-----KCSVQKAGTTFTHKVNLMDSVHILCSDDCFNQFRTSNKLNNGSCMNCGGICY---GTDAPCQSLQI	328

S1254T

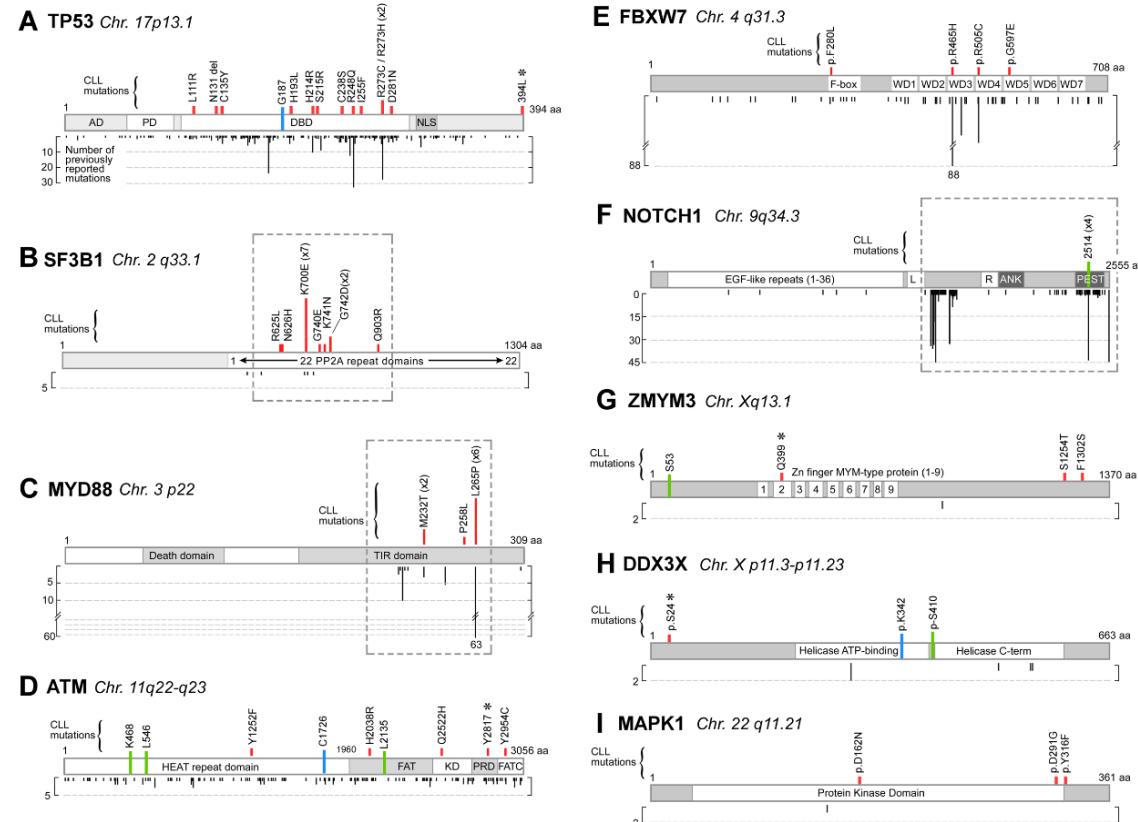
Human	1213	MFFNTKFFGLQTAEEHMQLSFTNVVRQSRKCTTPRGTTKVVSIRYYAPVRQRKGRDTGPGKRKE-----DEAPILEQRE	1287
Mouse	1214	MFFNTKFFGLQTAEEHMQLSFTNVVRQSRKCTTPRGTTKVVSIRYYAPVRQRKGRDTGPGKRKE-----DET-ILEQRE	1287
Zebrafish	1361	IYFFTQYNYRTAEQHRLLSFGHIVR-----CSRSKGNTKVACLRFYPPKDEADGV---PAKRRKEEGEEEDET-VYEIKE	1432

F1302S

Human	1288	NRMNPLRCPVKFYEFYLSKCPCESLRTRNDVFYLQPERSCIAESPLWYSVIPMDRSMLESMLNRILAVREIY---EELGRP	1364
Mouse	1288	NRMNPLRCPVKFYEFYLSKCPCESLRTRNDVFYLQPERSCIAESPLWYSVIPMDRSMLESMLNRILAVREIY---EELGRP	1364
Zebrafish	1433	NSDNPLRCPVRLYEFYLSKCSPSVRQRTDFYLSPERSCVPNSPMWFISALSDEALNSMLTRILTVERLHLDTEKTPAD	1512

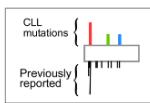
### **Supplemental Figure 3. Mutation types and locations in the 9 significantly mutated**

**genes.** **A-I.** Type (missense, splice-site, nonsense) and location of mutations in the 9 significantly mutated genes discovered among the 91 CLL samples (top) compared to previously reported mutations in literature or in the COSMIC database (v76) (bottom). Dashed boxes in **B**, **C** and **F** indicate mutations localizing to a discrete gene territory.



### Key and Notes:

| Missense  
| Splice  
| Frame shift  
\* Nonsense



**A**  
 AD = Activation domain (1-63 aa)  
 PD = Proline-rich domain (64-92 aa)  
 DBD = DNA binding domain (100-300 aa)  
 NLS = Nuclear localization  
 signaling domain (316-325 aa)

Topological domain (Extracellular): (18-1735 aa)  
 Transmembrane domain: (1736-1756 aa)  
 Topological domain (Cytoplasmic): (1757-2555 aa)  
 EGF-like repeats (1-36): (20-1426 aa)  
 L repeats (1-3): (1449-1571 aa)

C

	Zinc finger MYM-type 1	(332 - 366 aa)
)	Zinc finger MYM-type 2	(378 - 422 aa)
aa)	Zinc finger MYM-type 3	(429 - 464 aa)
	Zinc finger MYM-type 4	(477 - 511 aa)
	Zinc finger MYM-type 5	(521 - 559 aa)
	Zinc finger MYM-type 6	(567 - 604 aa)
	Zinc finger MYM-type 7	(612 - 646 aa)
	Zinc finger MYM-type 8	(653 - 692 aa)
	Zinc finger MYM-type 9	(699 - 733 aa)

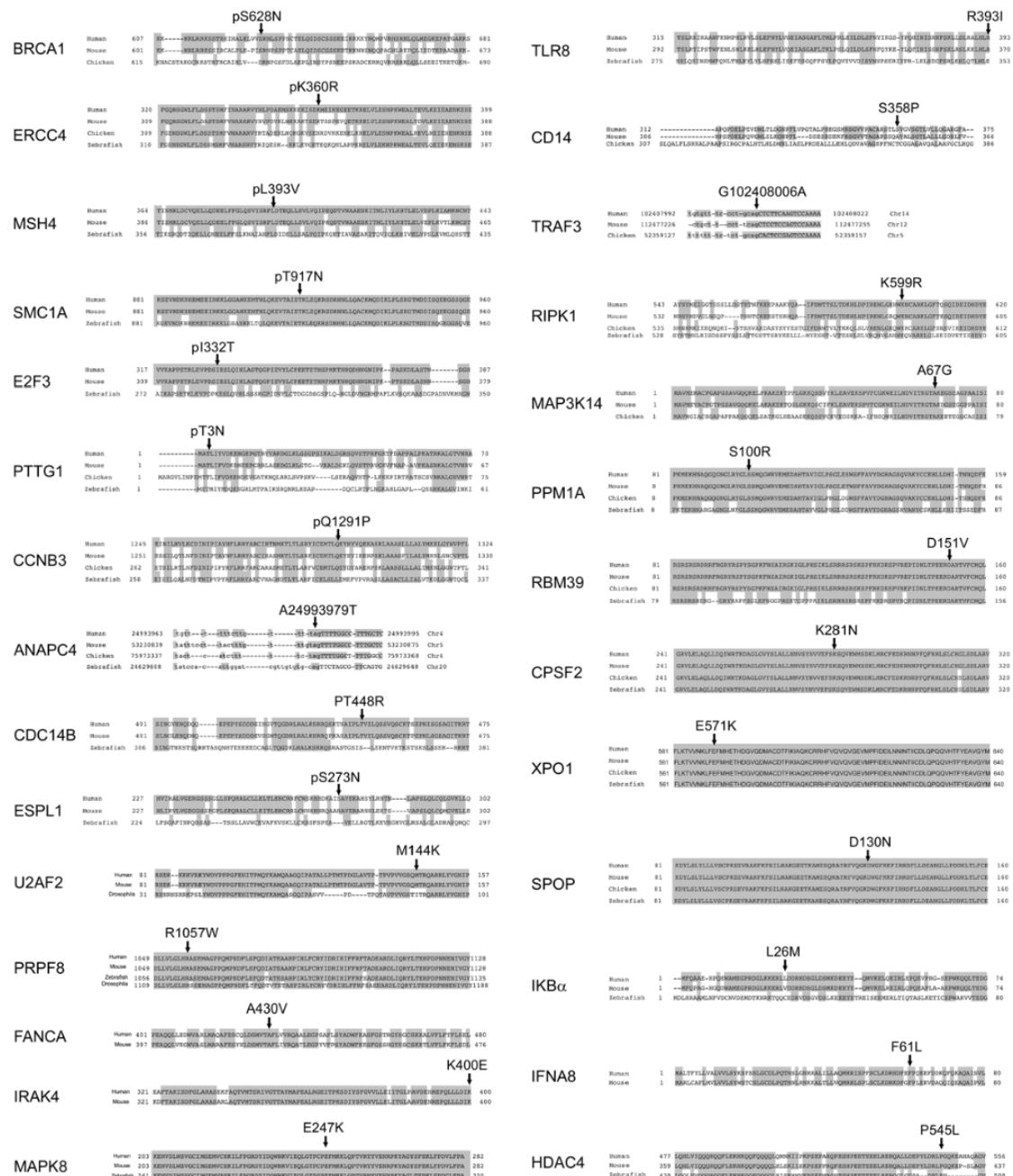
D

PRD = PIKK-regulatory domain (2961-3025  
 FATC = FAT-c-term domain (3024-3056 aa)

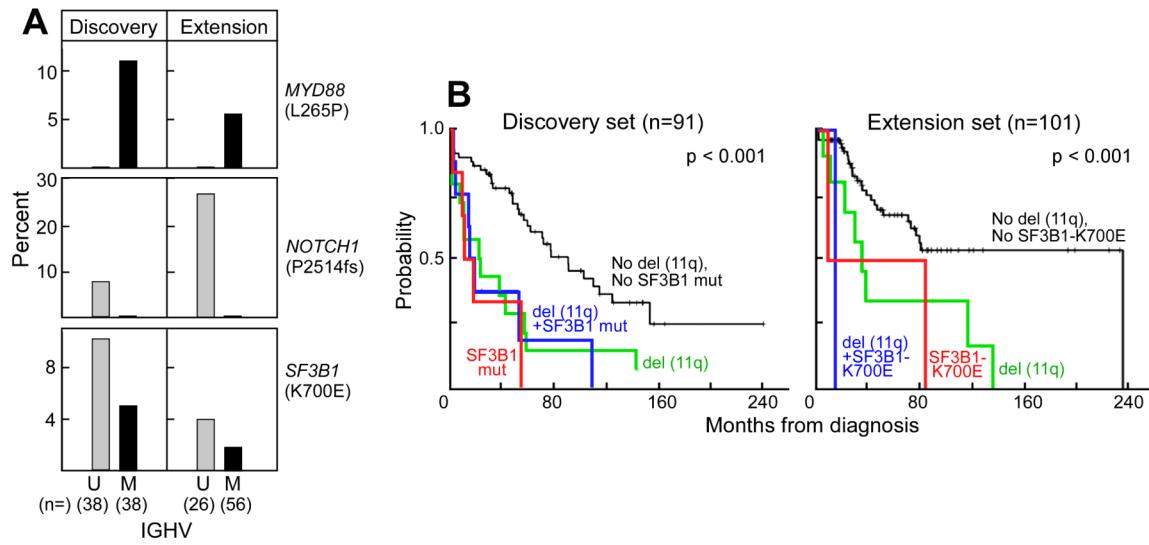
**E**  
 F-box domain (278-324 aa) WD4 = (500-536 aa)  
 WD1 = (378-418 aa) WD5 = (539-578 aa)  
 WD2 = (420-456 aa) WD6 = (580-618 aa)  
 WD3 = (459-498 aa) WD7 = (622-659 aa)

Protein Kinase Domain (25-313 aa)

**Supplemental Figure 4. Mutations in genes that are pathway related to driver mutations occur in evolutionarily conserved locations.** Where available, alignments of gene sequences around each mutation are shown for human, mouse, chicken and zebrafish, genes (USCS Genomic Bioinformatics: <http://genome.ucsc.edu>).

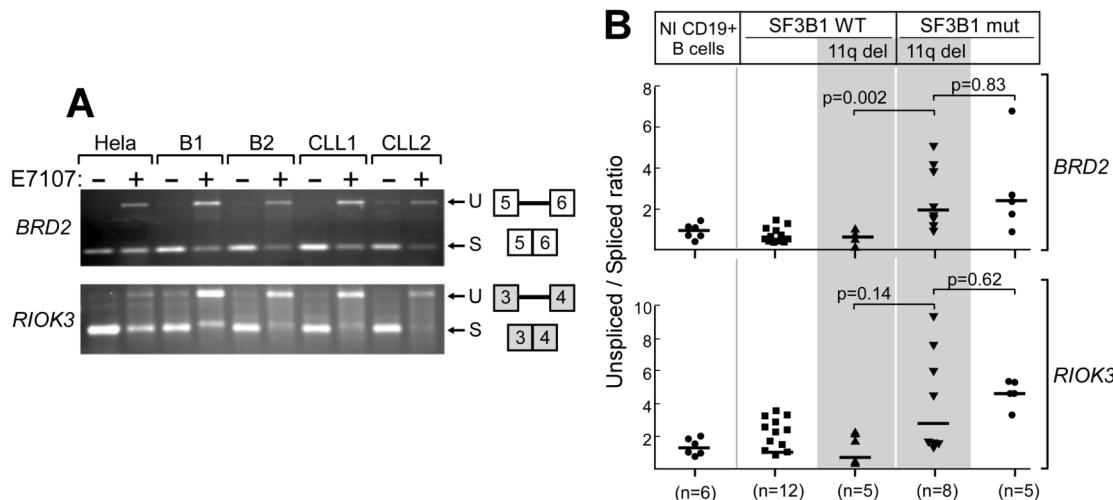


**Supplemental Figure 5. Mutation in *SF3B1* is associated with earlier TTFT.** **A.** Percent samples harboring the *SF3B1*-K700E, *MYD88*-L265P or *NOTCH1*-P2514fs mutations, within the 78 exomes with known IGHV mutation status (U-unmutated; M-mutated), and the 82 extension set CLL samples with known IGHV mutation status. Mutations were detected by exome sequencing for the 78 samples in the discovery set and by Mass Sequenom genotyping for the 82 samples analyzed in the extension set. **B.** Kaplan-Meier curves of the probability of time-to-first-therapy for 91 patients included in our discovery set (left), and for 101 patient samples that underwent genotyping of the *SF3B1*-K700E mutation in the extension set (right). Samples were categorized based on the presence or absence of del(11q) and the presence or absence of *SF3B1* mutations. Patients with either del(11q) or *SF3B1* mutation or both demonstrate significantly shorter time to first therapy as compared to all others (log-rank test).

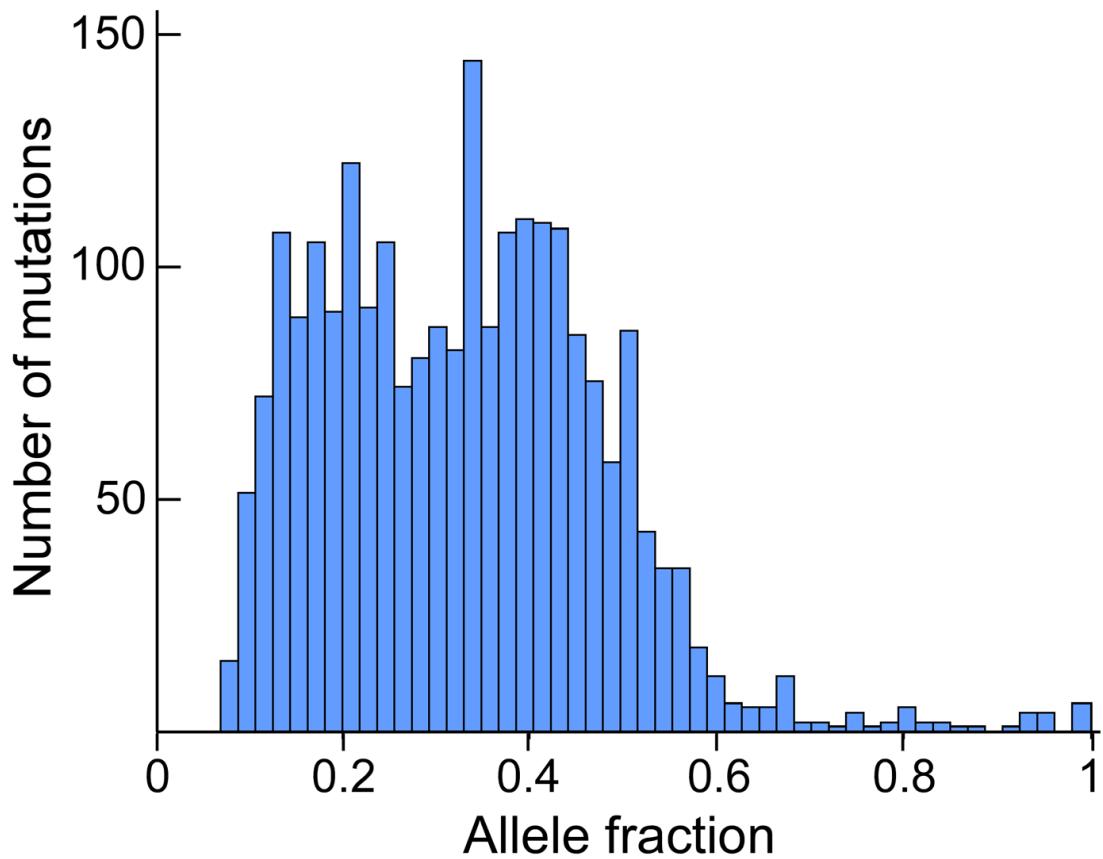


**Supplemental Figure 6. Altered splicing in CLL is associated with mutation in *SF3B1***

**A.** Treatment with E7107, which targets the SF3b complex generates increased ratio of unspliced to spliced *RIOK3* and *BRD2* mRNA. Hela cells, normal CD19+ B cells and CLL cells were treated with E7107 for 4 hours. Unspliced (U) and spliced (S) *BRD2* and *RIOK3* were amplified by reverse transcription PCR and analyzed by agarose gel electrophoresis. **B.** The relative amounts of spliced and unspliced *BRD2* and *RIOK3* mRNAs, measured by quantitative PCR, based on presence or absence of del(11q) and WT or mut *SF3B1* are shown. The ratios of unspliced to spliced mRNAs were normalized to the percentage of leukemia cells per sample, and comparisons were calculated using the Wilcoxon rank sum test.



**Supplemental Figure 7.** Distribution of allelic fraction of 2348 coding mutations (535 synonymous, 1813 non-synonymous) detected from 91 sequenced CLL samples.



**Table 1:** A complete list of somatic non-synonymous mutations in the final analysis set of 3 CLL genomes and 88 CLL exomes. Genomic position and transcript annotation are provided with respect to genome build hg18 (<http://genome.ucsc.edu>).

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
APEX2	27301	55045451	Missense	c.360C>G	p.A95G	uc004dtz.1	P1
ASXL1	171023	30488000	Nonsense	c.4250C>G	p.S1275*	uc002wxs.1	P1
ATP13A2	23400	17196202	Missense	c.1129T>G	p.C365W	uc001baa.1	P1
BZRAP1	9256	53745004	Missense	c.3048C>T	p.S726F	uc002ivx.2	P1
C11orf61	79684	124175119	Missense	c.391A>G	p.E123G	uc001qba.1	P1
C7orf51	222950	99924946	Missense	c.1825G>A	p.A556T	uc003uvd.1	P1
CREB3L2	64764	137263565	Missense	c.585A>G	p.M64V	uc003vtw.1	P1
DNMT3L	29947	44493352	Missense	c.1464T>C	p.I327T	uc002zeh.1	P1
GGA1	26088	36358654	Missense	c.2275G>T	p.G637V	uc003atc.1	P1
HIPK2	28996	138908403	Missense	c.3581A>C	p.Y1136S	uc003vvf.2	P1
INPP4B	8821	143263948	Missense	c.2559A>T	p.Q655L	uc003iix.2	P1
MAPK8	5599	49303987	Missense	c.963G>A	p.E247K	uc009xnz.1	P1
MYO10	4651	16756173	Missense	c.2839G>C	p.A791P	uc003jft.2	P1
R3HDM2	22864	55936537	Missense	c.2865G>C	p.G825A	uc003snt.2	P1
SLIT2	9353	20159235	Missense	c.2576C>T	p.T791M	uc003gpr.1	P1
TMEM51	55092	15418430	Missense	c.914T>A	p.D122E	uc001awv.2	P1
TOLLIP	54472	1273536	Missense	c.209T>G	p.V33G	uc001lte.1	P1
TSFM	10102	56476508	Missense	c.965T>C	p.S306P	uc001sqh.2	P1
UROC1	131669	127707353	Missense	c.726C>T	p.R232W	uc010hs.1	P1
ZFR2	23217	3759936	Frame_Shift_Ins	c.2490_2491insG	p.G826fs	uc002lyw.2	P1
ZNF536	9745	35731163	Missense	c.2935G>A	p.E933K	uc002nsu.1	P1
ZNF578	147660	57705665	Missense	c.463G>T	p.E73D	uc002ppz.2	P1
ADAMTSL3	57188	82476242	Missense	c.4484A>C	p.E1420D	uc002bjz.2	P2
ARHGEF10L	55160	17894135	Frame_Shift_Del	c.1007_1022delTT	p.F51fs	uc001bas.1	P2
C14orf37	145407	57674770	Missense	c.1171G>A	p.E354K	uc001xdc.1	P2
C4orf22	255119	82010250	Missense	c.513C>T	p.T155M	uc010ijp.1	P2
CPSF2	53981	91678442	Missense	c.1080G>T	p.K281N	uc001yah.1	P2
DMC1	11144	37265361	Missense	c.672G>A	p.R166H	uc003avz.1	P2
EHBP1L1	254102	65114138	Missense	c.4329G>A	p.R1355Q	uc001oeo.2	P2
GPR61	83873	109887249	Missense	c.765G>T	p.A28S	uc001dxy.2	P2
GRIP2	80852	14556888	Missense	c.223A>G	p.R75G	uc003byt.1	P2
KIAA1244	57221	138625638	Missense	c.1325A>G	p.Q442R	uc003qhu.2	P2
MAK	4117	10872696	Missense	c.2076T>G	p.W616G	uc003mzl.1	P2
MORC3	23515	36654161	Missense	c.1304G>A	p.C416Y	uc002yvi.1	P2
MYOM1	8736	3145015	Missense	c.1907T>G	p.Y525D	uc002klp.1	P2
NAIF1	203245	129868759	Missense	c.454C>A	p.T148K	uc004bta.1	P2
NBPF16	728936	147019954	Frame_Shift_Del	c.1538_1544delTT	p.D449fs	uc001esf.2	P2
NET1	10276	5486369	Frame_Shift_Del	c.1048_1066delCT	p.L304fs	uc001iia.1	P2
NSL1	25936	211024336	Nonsense	c.470G>T	p.E146*	uc001hjn.1	P2
PCDHGB4	8641	140749175	Missense	c.1540G>A	p.A514T	uc003lk.1	P2
PIGX	54965	197939992	Missense	c.713A>T	p.R144S	uc010iaj.1	P2
RP1	6101	55700154	Missense	c.1307T>C	p.F387L	uc003xsd.1	P2
RSPO4	343637	892700	Missense	c.570G>A	p.G158D	uc002wej.1	P2
SKI	6497	2150476	Frame_Shift_Del	c.483_484delGC	p.Q137fs	uc001aja.2	P2
SLC2A14	144195	7861773	Missense	c.2058G>C	p.R422P	uc001qtk.1	P2
TARSL2	123283	100082062	Nonsense	c.107C>T	p.Q18*	uc002bxm.1	P2
TNNT3	7140	1916276	Missense	c.967A>T	p.K252I	uc001luu.2	P2
TRAF7	84231	2160615	Splice_Site_Ins	c.e5_splice_site		uc002cow.1	P2
TRIM7	81786	180554912	Frame_Shift_Ins	c.1462_1463insA	p.L465fs	uc003mmz.1	P2
ZNF296	162979	50267276	Missense	c.908T>G	p.V284G	uc002pao.1	P2
ZNF462	58499	108730641	Missense	c.4916G>A	p.V1543M	uc004bcz.1	P2
BAZ2A	11176	55289786	Splice_Site_SNP	c.e10_splice_site		uc001slq.1	P3
CADPS2	93664	121901798	Missense	c.2034G>A	p.R624H	uc010lkp.1	P3
CENPE	1062	104251549	Missense	c.7699G>A	p.V2537I	uc003hx.1	P3
DCLK1	9201	35295012	Nonsense	c.1620G>T	p.G470W	uc001uvf.1	P3
DDX3X	1654	41081630	Nonsense	c.926C>A	p.S24*	uc004dfe.1	P3
DNA2	1763	69901564	Missense	c.322C>G	p.P108A	uc001jof.1	P3
EOMES	8320	27734163	Missense	c.1520G>A	p.R507H	uc003cdy.2	P3
F9	2158	138446978	Missense	c.261T>G	p.F78V	uc004fas.1	P3
IFI16	3428	157288330	Frame_Shift_Del	c.2025_2026delTA	p.Y579fs	uc001ftg.1	P3
MYH1	4619	10353626	Missense	c.1582G>T	p.M496I	uc002gmo.1	P3
PLCL1	5334	198656746	De_novo_Start_OutOfFrame	c.146G>A		uc002uuw.2	P3
PPP1CC	5501	109643278	Nonsense	c.1112C>T	p.Q320*	uc001tru.1	P3
PRICKLE1	144165	41149628	Missense	c.505A>T	p.E92V	uc001rnl.1	P3
PTPRT	11122	40177338	Missense	c.3255C>T	p.T1024M	uc010ggj.1	P3
RFX7	64864	54174766	Frame_Shift_Del	c.2451_2452delGA	p.E817fs	uc010bf.1	P3
SERPINB2	5055	59721264	Missense	c.1065C>A	p.D331E	uc002ljo.1	P3
TP53	7157	7518263	Missense	c.937G>A	p.R248Q	uc002gim.2	P3
ANKRD30A	91074	37459205	Missense	c.334G>A	p.V79I	uc001iza.1	P4
ATXN7L3	56970	39630295	Splice_Site_SNP	c.e3_splice_site		uc002ifz.1	P4
C15orf59	388135	71819930	Missense	c.608G>A	p.G88D	uc002avy.1	P4
CPVL	54504	29070353	Missense	c.1105A>T	p.Y329F	uc003szv.1	P4
DAB1	1600	57249009	Missense	c.2289G>A	p.E539K	uc001cys.1	P4
DES	1674	219993578	Missense	c.939G>A	p.A285T	uc002vll.1	P4

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
HERPUD1	9709	55533552	Missense	c.1322G>A	p.V305I	uc002eke.1	P4
HFM1	164045	91618348	Missense	c.1007G>A	p.A303T	uc001doa.2	P4
KCNJ2	3759	65683052	Missense	c.678G>A	p.V93I	uc010dfg.1	P4
MAVS	57506	3793248	Missense	c.1140C>T	p.S324F	uc002wjw.2	P4
NLGN3	54413	70306007	Missense	c.2126G>A	p.V608M	uc004dzb.1	P4
OR6A2	8590	6772980	Missense	c.736T>C	p.I179T	uc001mes.1	P4
PPFIBP1	8496	27708589	Missense	c.1371T>C	p.C332R	uc001ric.1	P4
RIN2	54453	19918809	Missense	c.1958T>G	p.V641G	uc002wro.1	P4
SPAG8	26206	35800295	Nonsense	c.1327C>A	p.Y404*	uc003zye.1	P4
ARHGEF10	9639	1812236	Missense	c.950G>A	p.E258K	uc003wpr.1	P5
ATAD3B	83858	1413149	Missense	c.1359C>G	p.R420G	uc001aif.1	P5
ATM	472	107741029	Missense	c.9246A>G	p.Y2954C	uc001pkb.1	P5
C12orf48	55010	101113976	Missense	c.1737A>C	p.K425T	uc001tjg.1	P5
CCDC18	343099	93492662	Missense	c.3767G>A	p.R1200Q	uc001dpq.1	P5
FMNL3	91010	48342029	Nonsense	c.673C>T	p.Q147*	uc001ruv.1	P5
KCNJ5	3762	128286871	Missense	c.807A>T	p.I165F	uc001qet.1	P5
KCNJ6	3763	38008528	Missense	c.1339G>A	p.D268N	uc002ywo.1	P5
KDR	3791	55659683	Missense	c.2614A>G	p.T771A	uc003has.1	P5
LCP1	3936	45631039	Nonsense	c.277C>T	p.R51*	uc001vaz.2	P5
MED27	9442	133944883	Missense	c.192A>T	p.Q57L	uc004cbe.1	P5
MTOR	2475	11110752	Missense	c.6008A>T	p.T1977S	uc001asd.1	P5
MUC6	4588	1009308	Missense	c.4048A>C	p.T1333P	uc001lsw.2	P5
MYD88	4615	38157645	Missense	c.794T>C	p.L265P	NM_002468	P5
PCDH17	27253	57106480	Missense	c.2691A>T	p.N600I	uc001vhq.1	P5
PHLPP2	23035	70267992	Missense	c.1336G>A	p.V444M	uc002fax.1	P5
PRKCQ	5588	6580493	Missense	c.596G>T	p.G171V	uc001iji.1	P5
RALYL	138046	85604230	Missense	c.292C>A	p.A53D	uc003yct.2	P5
ROS1	6098	117780921	Missense	c.4445G>A	p.A1416T	uc003pxp.1	P5
SIM1	6492	101002763	Missense	c.1037T>C	p.L277P	uc003pj.2	P5
SVEP1	79987	112291768	Missense	c.2250T>C	p.F638S	uc010mtz.1	P5
ZNHIT6	54680	85940432	Missense	c.1149A>G	p.K339E	uc001dlh.1	P5
CCDC67	159989	92736975	Missense	c.399T>C	p.F100S	uc001pdq.1	P6
CCDC94	55702	4218759	Frame_Shift_Ins	c.880_881insC	p.A283fs	uc002lvz.2	P6
CFH	3075	194964125	Missense	c.2503T>C	p.S755P	uc001gtj.2	P6
COL14A1	7373	121332172	Missense	c.3003G>T	p.G913V	uc003yox.1	P6
DDX3X	1654	41089376	Splice_Site_SNP	c.e11_splice_site	uc004dfe.1		
FERMT1	55612	6048118	De_novo_Start_OutOfFrame	c.873C>T	uc010gbt.1		
MTCH1	23787	37053843	Missense	c.580G>T	p.V194F	uc003one.2	P6
MYCBP2	23077	76540862	Missense	c.11987G>A	p.D3966N	uc001vkf.1	P6
MYO7A	4647	76573419	Splice_Site_Del	c.e27_splice_site	uc009yur.1		
OR2S2	56656	35947816	Missense	c.336T>C	p.S84P	uc003yzt.2	P6
POU6F2	11281	39466752	Missense	c.1526G>A	p.R495H	uc003thb.1	P6
SF3B1	23451	197975726	Missense	c.1924A>C	p.N626H	uc002uu.1	P6
SMAD1	4086	146655259	Missense	c.460A>G	p.K15R	uc003ikc.1	P6
SPATA6	54558	48649798	Missense	c.495T>A	p.F110L	uc001crr.1	P6
ZNF492	57615	22639513	Missense	c.1333C>T	p.A401V	uc002nqw.2	P6
CCNY	219771	35881993	Missense	c.800T>C	p.I207T	uc001iyw.2	P7
COL28A1	340267	7364940	Missense	c.3344T>C	p.L1076S	uc003src.1	P7
DNAJB2	3300	219857865	Frame_Shift_Ins	c.1124_1125insG	p.L296fs	uc002vkx.1	P7
EIF4A3	9775	75725883	Missense	c.1058A>G	p.T294A	uc002xjs.1	P7
ELF5	2001	34458369	Missense	c.1000C>T	p.A257V	uc001mvo.1	P7
GCNT3	9245	57698729	Missense	c.1590G>A	p.A334T	uc002agd.1	P7
IGFBP3	3486	45922781	Missense	c.791G>A	p.R220H	uc003trr.1	P7
LAMA2	3908	129517441	Missense	c.1231G>A	p.G376S	uc003qbn.1	P7
MBTPS2	51360	21810543	Nonsense	c.1508G>A	p.W470*	uc004dac.1	P7
MYLK3	91807	45320522	Missense	c.1803A>T	p.I563F	uc002eei.2	P7
MYOC	4653	169888292	Nonsense	c.105G>A	p.W28*	uc001ghu.1	P7
ONECUT2	9480	53254407	Missense	c.493T>C	p.L154P	uc002igo.1	P7
PAMR1	25891	35410637	Missense	c.2100C>T	p.A686V	uc001mwf.1	P7
PCDHA10	56139	140217127	Missense	c.1310C>G	p.T437R	uc003lhx.1	P7
PCDHGB3	56102	140731583	Missense	c.1438G>A	p.D480N	uc003ljw.1	P7
POT1	25913	124290777	Missense	c.1010C>T	p.R137C	uc003vlm.1	P7
RARS	5917	167866405	Missense	c.1378G>A	p.G446E	uc003lzx.1	P7
SPIRE1	56907	12496637	Nonsense	c.858C>T	p.R271*	uc002kre.1	P7
TMC2	117532	2523528	Missense	c.1006G>A	p.G331R	uc002wgf.1	P7
ZDBF2	57683	206881135	Missense	c.3888G>A	p.R1213Q	uc002vbp.2	P7
ASH2L	9070	38082335	Missense	c.168C>T	p.A37V	uc003xkt.2	P8
ATM	472	107695947	Frame_Shift_Del	c.6789_6789delT	p.L2135fs	uc001pkb.1	P8
COL22A1	169044	139728095	Missense	c.3907C>G	p.P1154A	uc003yyd.1	P8
DMXL2	23312	49582517	Missense	c.2995G>A	p.A924T	uc002abf.1	P8
DYRK1A	1859	37784464	Missense	c.857T>G	p.L261R	uc002yw.1	P8
GADL1	339896	30817419	Missense	c.1263G>C	p.E406Q	uc003ceq.1	P8
GNB1	2782	1727802	Missense	c.571T>C	p.I80T	uc001aif.1	P8
GRID2	2895	94909513	Missense	c.2748C>A	p.S830R	uc003hsz.2	P8

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
HPS5	11234	18290111	Missense	c.423T>G	p.L49V	uc001mod.1	P8
ITGA5	3678	53099099	Frame_Shift_Del	c.213_219delCCA	p.P49fs	uc001sga.1	P8
LILRA4	23547	59541523	Missense	c.368C>T	p.A104V	uc002qfj.1	P8
MAMD2	256691	71936324	Missense	c.1564C>T	p.P324S	uc004ahm.1	P8
SF3B1	23451	197974856	Missense	c.2273G>A	p.G742D	uc002uee.1	P8
TMPRSS9	360200	2356419	Missense	c.616G>T	p.G206C	uc002lvw.1	P8
ANKRD26	22852	27358293	Splice_Site_SNP	c.e26_splice_site		uc009xku.1	P9
BCR	613	21853993	Missense	c.1442C>G	p.I282M	uc002zww.1	P9
CBARA1	10367	73937975	Missense	c.735G>A	p.G201E	uc001jt8.1	P9
CD14	929	139991681	Missense	c.1426T>C	p.S358P	uc003lgi.1	P9
DIS3	22894	72245834	Nonsense	c.1602A>T	p.R410*	uc001vix.2	P9
GFB1	8729	104129636	Missense	c.5050A>T	p.I1604F	uc001kux.1	P9
GJB2	2706	19661627	Missense	c.309C>T	p.R32C	uc001umy.1	P9
GNB2	2783	100113730	Missense	c.829T>G	p.S191A	uc003uw.1	P9
HECTD1	25831	30712649	Missense	c.1207A>G	p.M240V	uc001wrc.1	P9
IGSF22	283284	18695022	Missense	c.1265G>T	p.V359L	uc009yht.1	P9
IQGAP1	8826	88785740	Splice_Site_SNP	c.e8_splice_site		uc002bp1.1	P9
MED12	9968	70256023	Missense	c.374G>C	p.A59P	uc004ddy.1	P9
MMP16	4325	89200142	Missense	c.1056T>A	p.N258K	uc003yeb.2	P9
PLSCR1	5359	147722532	Splice_Site_SNP	c.e6_splice_site		uc003evx.2	P9
REV1	51455	99388904	Missense	c.2923C>T	p.T904I	uc002tad.1	P9
RHO	6010	130734178	Missense	c.904G>A	p.S270N	uc003emt.1	P9
SH3BP4	23677	235627037	Nonsense	c.3123C>G	p.Y910*	uc002zvp.1	P9
SLC7A4	6545	19715741	Missense	c.429A>G	p.N121D	uc002zud.1	P9
SNX19	399979	130255889	Missense	c.3144A>G	p.N866D	uc001gk.2	P9
TET1	80312	70074858	Missense	c.2871A>T	p.N789I	uc001jok.2	P9
TP53	7157	7518243	Missense	c.957A>T	p.I255F	uc002gim.2	P9
TTC7A	57217	47127944	Frame_Shift_Del	c.2323_2323delA	p.Q652fs	uc010ffb.1	P9
UBR5	51366	10385535	Missense	c.2899C>G	p.L956V	uc003ykr.1	P9
ZSCAN18	65982	63292018	Splice_Site_SNP	c.e3_splice_site		uc002qrh.1	P9
CELSR2	1952	109594496	Missense	c.333G>A	p.R91K	uc001dxa.2	P10
CEMP1	752014	2520913	Missense	c.519A>G	p.K55E	uc002cqr.2	P10
FAM155B	27112	68666141	Missense	c.1084T>G	p.L346V	uc004dkx.1	P10
FAT4	79633	126592681	Missense	c.11060A>G	p.D3687G	uc003ifj.2	P10
HSPA4L	22824	128946323	Missense	c.1422G>A	p.R390H	uc003ifm.1	P10
LRRC56	115399	541685	Frame_Shift_Ins	c.1320_1321insT	p.D277fs	uc001lpw.1	P10
MET	4233	116126605	Missense	c.418C>A	p.D77E	uc010lkh.1	P10
MYL5	4636	664336	Missense	c.436A>C	p.M111L	uc003gav.1	P10
NTN3	4917	2463275	Missense	c.1476C>T	p.P425S	uc002cqj.1	P10
PRKCI	5584	171496391	Splice_Site_SNP	c.e15_splice_site		uc003fgs.2	P10
TMRSS6	164656	35794601	Splice_Site_SNP	c.e17_splice_site		uc003aqt.1	P10
UBA1	7317	46958727	Missense	c.3047A>G	p.N966D	uc004dhj.2	P10
WDFY3	23001	85920389	Missense	c.4669G>A	p.A1421T	uc003hpd.1	P10
ZNF423	23090	48227712	Missense	c.3150C>T	p.T951M	uc002efs.1	P10
CDH23	64072	73170595	Missense	c.4876C>T	p.S1500F	uc001jrx.2	P10
DIS3	22894	72235744	Missense	c.2347A>G	p.E658G	uc001vix.2	P10
DSCAML1	57453	116897252	Missense	c.1198C>T	p.T399M	uc001prh.1	P11
GDF15	9518	18360107	Missense	c.321T>G	p.S97A	uc002niv.2	P11
HCFC1R1	54985	3013266	Frame_Shift_Ins	c.382_383insC	p.P83fs	uc002csx.1	P11
HK3	3101	176248421	Missense	c.1039T>G	p.V322G	uc003rma.1	P11
LOXL4	84171	100010861	Missense	c.621A>G	p.E157G	uc001kpa.1	P11
MST1	4485	49699802	Missense	c.440A>C	p.K143Q	uc003cxg.1	P11
NIPA1	123606	20612340	Missense	c.258T>G	p.V78G	uc001yvc.1	P11
NME6	10201	48315016	Missense	c.65A>G	p.S7G	uc003csco.1	P11
PTGIR	5739	51816468	Missense	c.1183T>G	p.V357G	uc002pex.1	P11
RUND C3B	154661	87167736	Missense	c.762G>T	p.C118F	uc003ujb.1	P11
SALL4	57167	49841374	Missense	c.1156C>T	p.A352V	uc002xwh.2	P11
SPTB	6710	64323005	Missense	c.3485A>G	p.E1144G	uc001xhr.1	P11
STARD13	90627	32585045	Missense	c.2424C>G	p.Q769E	uc001uuw.1	P11
TAS1R2	80834	19039411	Missense	c.1790C>T	p.R597C	uc001bba.1	P11
ATRX	546	76794441	Frame_Shift_Ins	c.4607_4608insC	p.E1459fs	uc004ecp.2	P12
CXorf22	170063	35898921	Missense	c.1989T>A	p.Y644N	uc004ddj.1	P12
DZIP1L	199221	139273352	Missense	c.1801T>C	p.S480P	uc003erq.1	P12
ELMOD2	255520	141678014	Splice_Site_SNP	c.e5_splice_site		uc003iih.1	P12
FAM47A	158724	34059355	Missense	c.995C>T	p.P321L	uc004ddg.1	P12
FBXW7	55294	153466739	Missense	c.1662C>T	p.R505C	uc003ims.1	P12
GALNT13	114805	154806955	Missense	c.582G>T	p.D160Y	uc002tyt.2	P12
ITIH2	3698	7812013	Missense	c.1534G>A	p.D458N	uc001ijs.1	P12
KCNA2	3737	110948749	Missense	c.675G>A	p.G60E	uc001dzu.1	P12
LTB	4050	31657349	Missense	c.208T>C	p.I67T	uc003nul.1	P12
MLL5	55904	104534235	Missense	c.3161T>G	p.F876C	uc003vcm.1	P12
MRPS14	63931	173259164	Missense	c.21G>A	p.A2T	uc001gkk.1	P12
NAV2	89797	20023535	Missense	c.4075T>A	p.D1238E	uc009yhw.1	P12
NOBOX	135935	143729428	Frame_Shift_Ins	c.487_488insC	p.R163fs	uc003wen.1	P12

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
NUDT9	53343	88575339	Missense	c.613T>C	p.V97A	uc003hqq.1	P12
SLTRK4	139065	142544118	Missense	c.284G>A	p.L825I	uc004fbx.1	P12
SUV420H1	51111	67695088	Missense	c.1203A>G	p.N316S	uc001onm.1	P12
TRHDE	29953	71343212	Missense	c.3141C>A	p.F1015L	uc001sxa.1	P12
CCDC99	54908	168960894	Missense	c.1636C>T	p.R453C	uc003mae.2	P13
CELSR2	1952	109615413	Missense	c.7709C>T	p.R2550W	uc001dxa.2	P13
DNTTIP1	116092	43854757	Missense	c.208T>G	p.V47G	uc002xpk.1	P13
EEF1D	1936	144733919	Missense	c.1989C>T	p.A587V	uc003yyq.1	P13
EGF	1950	111151823	Missense	c.993T>C	p.L251P	uc010imk.1	P13
HGD1C	613227	49650547	Frame_Shift_Ins	c.285_286insA	p.S95fs	uc009zlu.1	P13
KIAA2022	340533	73876738					
KRT5	3852	51200162	Missense	c.4693A>G	p.E1460G	uc004ebj.1	P13
MAOA	4128	43456087	Missense	c.349C>G	p.S62R	uc001san.1	P13
MPEG1	219972	58736287	Missense	c.512G>A	p.A111T	uc004dfy.1	P13
NISCH	11188	52499853	Missense	c.784G>A	p.D210N	uc001nnu.2	P13
POLA1	5422	24645622	Missense	c.3840A>G	p.N1236D	uc003ded.2	P13
PTX3	5806	158643184	Missense	c.918A>G	p.S299G	uc004dbl.1	P13
RFX7	64864	54175584	Missense	c.1011G>A	p.A290T	uc003fbl.2	P13
SDCCAG3	10807	138418948	Frame_Shift_Ins	c.1228_1229insT	p.A341fs	uc004chi.1	P13
TAF1	6872	70519409					
TEKT1	83659	6644089	Missense	c.1850G>T	p.G600V	uc004dz.2	P13
TMEM8A	58986	362109	Missense	c.1348G>A	p.R413H	uc002gdt.1	P13
USF1	7391	159279072	De_novo_Start_OutOfFrame	c.2324G>A	p.S732N	uc002cgu.2	P13
ZC3H12B	340554	64633878					
ZMYM3	9203	70378786	Missense	c.266C>A	uc001fxj.1	P13	
ZNF253	56242	19863281	Splice_Site_SNP	c.e2_splice_site	uc010nko.1	P13	
ADPRHL1	113622	113146822					
C3orf59	151963	194000064	Missense	c.385G>T	p.D100Y	uc001vtq.1	P14
EML4	27436	42410840	Missense	c.602G>A	p.R92Q	uc003fsz.1	P14
FLNA	2316	153231043	Frame_Shift_Ins	c.3171C>A	p.P979T	uc002rsi.1	P14
KBTBD8	84541	67141034					
KIT	3815	55290365	Splice_Site_SNP	c.1280G>A	p.Q2546fs	uc004fk.2	P14
MATR3	9782	138689749					
MSH4	4438	76086496	Missense	c.2185G>T	p.A700S	uc003dmy.1	P14
NCOA4	8031	51250888	Missense	c.1218C>G	p.L393V	uc001dhd.1	P14
PRAMEF10	343071	12875552	Frame_Shift_Ins	c.1280G>A	p.G403R	uc001auo.1	P14
SIGLEC1	6614	3618723					
COL1A2	1278	93866333	Splice_Site_SNP	c.1711_1712insT	p.P1593fs	uc002wja.1	P14
CSMD1	64478	3598920					
KBTBD4	55709	47555943	Missense	c.1261C>T	p.R291*	uc001lrh.1	P15
PLK2	10769	57788768	Frame_Shift_Ins	c.975T>G	p.V87G	uc001nfw.1	P15
SAFB2	9667	5541342					
TBX4	9496	56912280	Missense	c.2683_2684insG	p.G824fs	uc002mcd.1	P15
TPST2	8459	25267466	Frame_Shift_Ins	c.1002T>A	p.I280N	uc010ddo.1	P15
TRAF3	7187	102408006					
ZAP70	7535	97707106	Splice_Site_SNP	c.1131_1132insT	p.L335fs	uc003jrn.1	P15
ACADSB	36	124789970					
CLCN3	1182	170854913	Splice_Site_SNP	c.3087G>A	p.I2006K	uc001lhb.1	P16
DLG5	9231	79251231					
EIF3E	3646	109316509	Splice_Site_SNP	c.3087G>A	p.R1006K	uc001jzk.1	P16
ELF4	2000	129035759					
FGFR1	53834	1008365	Missense	c.671G>T	p.E96*	uc003ymu.1	P16
FUBP1	8880	78205334	Frame_Shift_Ins	c.1146C>T	p.R329C	uc004evd.2	P16
GABRG3	2567	25446672					
HSPA8	3312	122435409	Missense	c.1711_1712insT	p.G110fs	uc001dil.1	P16
IDH1	3417	208816465	Missense	c.671G>T	p.A368T	uc001pyo.1	P16
MMD	23531	50836125	Splice_Site_SNP	c.875G>A	p.S210N	uc002vcs.1	P16
MTMR3	8897	28733305					
MUC16	94025	8950417	Missense	c.1202T>G	p.F292V	uc003agv.2	P16
NF1	4763	26565668	Missense	c.2602G>A	p.E800K	uc002mnp.1	P16
NOL11	25926	63166121	Missense	c.1799A>G	p.Y489C	uc002hgg.1	P16
NRCAM	4897	107623450	Missense	c.1873T>C	p.Y624H	uc002jgd.1	P16
OSBPL3	26031	24821349	Splice_Site_SNP	c.1925C>A	p.T485N	uc003vfb.1	P16
PAPPA	5069	118169807					
POLRMT	5442	581069	Missense	c.4939G>A	p.V1520M	uc004bjn.1	P16
PUM1	9698	31211608	Missense	c.349A>G	p.D98G	uc002lpf.1	P16
ZNF251	90987	145917948	Splice_Site_SNP	c.2133C>A	p.P668T	uc001bsk.1	P16
ABCB1	5243	87052934					
ATM	472	107660172	Missense	c.2162C>G	p.Q636E	uc003zdv.2	P16
BTAF1	9044	93746104	Splice_Site_SNP	c.4140A>T	p.Y1252F	uc003uiz.1	P17
DCBLD1	285761	117968864					
FAM123A	219287	24642073	Missense	c.1465C>T	p.S447L	uc003pxs.1	P17
FAT4	79633	126458429	Missense	c.1785C>T	p.P562L	uc001uqb.1	P17
GART	2618	33805432	Missense	c.1413T>G	p.H471Q	uc003if.2	P17
				c.2398G>C	p.E771Q	uc002yxr.1	P17

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
GPR126	57211	142756724	Splice_Site_SNP	c.e9_splice_site		uc010khe.1	P17
LRRC56	115399	541786	Frame_Shift_Del	c.1421_1421delA	p.E311fs	uc001lpw.1	P17
MYD88	4615	38157645	Missense	c.794T>C	p.L265P	NM_002468	P17
MYH9	4627	35011944	Missense	c.5247A>G	p.E1688G	uc003apg.1	P17
PKDCC	91461	42135942	Frame_Shift_Del	c.714_714delG	p.W177fs	uc002rsg.1	P17
SLC1A1	6505	4573063	Missense	c.1455G>A	p.G407R	uc003zij.1	P17
SLC6A16	28968	54505523	Missense	c.706T>G	p.F158V	uc002pmz.1	P17
USP10	9100	83336655	Missense	c.1209C>T	p.P356L	uc002ffl.1	P17
ZBTB11	27107	102866877	Missense	c.1474T>A	p.I415K	uc003dve.2	P17
ARHGAP30	257106	159287940	Missense	c.1554G>A	p.R403H	uc001fxl.1	P18
ATAD2B	54454	23896161	Missense	c.2521T>G	p.S743A	uc002rek.2	P18
BNC1	646	81723850	Missense	c.1245A>C	p.K386T	uc002bjt.1	P18
C1orf128	57095	23984842	Missense	c.535A>T	p.L137F	uc001bhq.1	P18
C1orf38	9473	28079147	Missense	c.669T>A	p.M214K	uc001bpc.2	P18
CDH9	1007	26941951	Missense	c.854A>G	p.R229G	uc003jgs.1	P18
DNAH10	196385	122899375	Missense	c.5766C>T	p.T1914M	uc001uft.2	P18
DNAH9	1770	11637610	Missense	c.8195T>G	p.H2709Q	uc002gne.1	P18
DOCK4	9732	111274412	Missense	c.2749G>A	p.R827Q	uc003vfy.1	P18
EMID2	136227	100877683	Splice_Site_SNP	c.e3_splice_site		uc003uyo.1	P18
ENPP1	5167	132227337	Splice_Site_SNP	c.e10_splice_site		uc003qcx.2	P18
FCER2	2208	7660294	Missense	c.929A>C	p.T251P	uc002mhm.1	P18
FLJ43860	389690	142552196	Missense	c.1886G>A	p.R602Q	uc003ywi.2	P18
GJA3	2700	19615309	Missense	c.291C>T	p.A40V	uc001umx.1	P18
GXYLT2	727936	73089128	Missense	c.911A>C	p.K304T	uc003dpg.1	P18
HMCN1	83872	184353212	Splice_Site_SNP	c.e77_splice_site		uc001grq.1	P18
IL26	55801	66905537	Missense	c.217T>A	p.I61K	uc001stx.1	P18
ITGB1	3688	33249301	Missense	c.1147A>T	p.I383F	uc001iwq.2	P18
ITGB1	3688	33251621	Missense	c.991A>T	p.I331F	uc001iwq.2	P18
KALRN	8997	125903617	Missense	c.8139T>G	p.F2680C	uc003ehg.1	P18
KLKB1	3818	187410194	Missense	c.1245G>A	p.V392I	uc003iy.1	P18
LPA	4018	160936387	Missense	c.3578C>G	p.S1153C	uc003sql.1	P18
MARK2	2011	63414276	Missense	c.369G>T	p.C16F	uc009yox.1	P18
MYD88	4615	38157263	Missense	c.695T>C	p.M232T	NM_002468	P18
OAT	4942	126090558	Missense	c.280T>C	p.L58S	uc001lh.p.2	P18
OMG	4974	26647400	Missense	c.264T>C	p.C26R	uc002hgi.1	P18
PCDH17	27253	57197163	Missense	c.4106T>G	p.L1072V	uc001vhq.1	P18
SETBP1	26040	40784471	Missense	c.1464G>A	p.A336T	uc010dn.i.1	P18
SLC12A5	57468	44102661	Splice_Site_SNP	c.e7_splice_site		uc002xr.b.1	P18
SLC8A1	6546	40196091	Missense	c.2752T>C	p.S910P	uc002rrx.1	P18
SSR1	6745	7246564	Missense	c.709A>G	p.N174S	uc003mx.2	P18
SULT1C3	442038	108238538	Missense	c.478G>C	p.D160H	uc002tdw.1	P18
TBCC	6903	42821345	Missense	c.518T>G	p.S149A	uc003osl.1	P18
TGM7	116179	41373040	Missense	c.97A>C	p.K31T	uc001zrf.1	P18
TSPAN19	144448	83937537	Missense	c.550C>T	p.T150I	uc009zsj.1	P18
XIRP2	129446	167809068	Missense	c.2938G>T	p.G974C	uc002udx.1	P18
ACOT2	10965	73106164	Missense	c.640T>G	p.V156G	uc001xon.2	P19
ADAM22	53616	87601578	Splice_Site_SNP	c.e12_splice_site		uc003ujp.1	P19
ANAPC4	29945	24993979	Splice_Site_SNP	c.e4_splice_site		uc003gro.1	P19
EPHB3	2049	185780358	Missense	c.2551G>T	p.R705L	uc003foz.1	P19
FAT4	79633	126589966	Missense	c.8345C>T	p.P2782L	uc003if.2	P19
GPRC6A	222545	117234665	Nonsense	c.918G>A	p.W299*	uc003pxj.1	P19
HYAL3	8372	50307803	Missense	c.508G>A	p.G79S	uc003czd.1	P19
M6PR	4074	8987663	Splice_Site_SNP	c.e4_splice_site		uc001qvf.1	P19
MAP3K14	9020	40723695	Missense	c.309C>G	p.A67G	uc002iiw.1	P19
METTL9	51108	21531465	Splice_Site_SNP	c.e2_splice_site		uc002dje.1	P19
MYCBP2	23077	76559647	Missense	c.10825T>A	p.N3578K	uc001vkf.1	P19
MYO3B	140469	170966437	Missense	c.2262G>A	p.E707K	uc002ufy.1	P19
PCLO	27445	82314427	Missense	c.14016G>A	p.S4576N	uc003uhx.2	P19
PDZD11	51248	69423689	Missense	c.732T>G	p.Y163D	uc004dye.1	P19
PIH1D1	55011	54642141	Nonsense	c.875G>A	p.W213*	uc002pns.1	P19
PPP1R12A	4659	78693829	Splice_Site_Del	c.e25_splice_site		uc001syz.1	P19
RAET1E	135250	150253673	Missense	c.118C>A	p.L20I	uc003ql.1	P19
RAI14	26064	34850443	Splice_Site_SNP	c.e14_splice_site		uc003jis.1	P19
SLC25A28	81894	101361042	Missense	c.778C>A	p.Q217K	uc001kp.x.2	P19
XKR8	55113	28165660	Missense	c.627G>T	p.A184S	uc001bph.1	P19
BAZ1A	11177	34334706	Missense	c.1713G>A	p.R382H	uc001wsk.1	P20
GPR133	283383	130017020	Missense	c.722C>T	p.H55Y	uc001uit.2	P20
IRF2	3660	185577718	Splice_Site_SNP	c.e3_splice_site		uc003iwf.2	P20
MUC5B	727897	1222539	Missense	c.7920C>T	p.A2621V	uc001ltb.2	P20
MYD88	4615	38157645	Missense	c.794T>C	p.L265P	NM_002468	P20
PA2G4	5036	54789956	Missense	c.1018C>A	p.T200N	uc001sjm.1	P20
PADI4	23569	17557919	Splice_Site_Ins	c.e14_splice_site		uc001baj.1	P20
PCDHAC1	56135	140287209	Missense	c.724C>A	p.P183Q	uc003lih.1	P20
WBSR17	64409	70523889	Missense	c.824T>C	p.I275T	uc003tv.y.1	P20

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WNT1	7471	47659762	Missense	c.547G>A	p.V117I	uc001rsu.1	P20
ABCA12	26154	215510478	Splice_Site_SNP	c.e51_splice_site		uc002vew.1	P21
AMBP	259	115863569	Missense	c.1072A>G	p.N270S	uc004bie.2	P21
ATP2A1	487	28821082	Missense	c.2582G>T	p.D800Y	uc002dro.1	P21
BEST1	7439	61484025	Missense	c.950C>T	p.P285L	uc001nsr.1	P21
BPHL	670	3068948	Missense	c.327A>G	p.T39A	uc003muy.1	P21
C4orf41	60684	184833316	Missense	c.842A>T	p.L222F	uc003ivx.1	P21
DGAT2L6	347516	69338638	Missense	c.743G>A	p.G216R	uc004dxx.1	P21
FRMD1	79981	168200785	Missense	c.1556C>A	p.H497Q	uc003qwo.2	P21
GATS	352954	99707409	Missense	c.141T>C	p.F45S	uc003uuu.2	P21
HSD3B2	3284	119766663	Missense	c.1789A>C	p.Y339S	uc001ehs.1	P21
HTT	3064	3116697	Missense	c.3238G>T	p.L1031F	uc010icr.1	P21
MOCS3	27304	49008917	Missense	c.148T>G	p.V44G	uc002xvy.1	P21
PFKFB1	5207	54992376	Missense	c.921G>A	p.A284T	uc004dty.1	P21
PRKRIR	5612	75741455	Missense	c.387T>A	p.H129Q	uc001oxh.1	P21
PTPN14	5784	212704727	Missense	c.314G>A	p.V15I	uc001hkk.1	P21
PTPRD	5789	8490768	Missense	c.2825G>T	p.R705L	uc003zk.1	P21
THBS1	7057	37666983	Missense	c.1443A>C	p.T422P	uc001zkh.1	P21
TMEM71	137835	133833342	Missense	c.328G>A	p.R62H	uc003ytp.1	P21
ULK2	9706	19625004	Missense	c.3145T>G	p.V882G	uc002gwm.2	P21
ALDH1L2	160428	103986645	Frame_Shift_Ins	c.597_598insG	p.P192fs	uc001ttc.1	P22
ANKRD49	54851	93871170	Missense	c.683G>A	p.A182T	uc001pew.1	P22
C15orf59	388135	71819444	In_frame_Del	c.1086_1094delCC	p.247_250SRHS>R	uc002avy.1	P22
CAD	790	27294389	Splice_Site_SNP	c.e2_splice_site		uc002rji.1	P22
CADM3	57863	157436261	Missense	c.1330T>G	p.F384C	uc001ftk.2	P22
CASC5	57082	38731489	Splice_Site_Del	c.e22_splice_site		uc010bbs.1	P22
CNOT6	57472	179926774	Frame_Shift_Del	c.1147_1147delG	p.K266fs	uc003mlx.1	P22
DGCR14	8220	17510249	Frame_Shift_Ins	c.330_331insAC	p.P98fs	uc002zou.1	P22
DUSP7	1849	52063271	Missense	c.584C>T	p.P175L	uc003dct.1	P22
EDEM3	80267	182929941	In_frame_Del	c.2910_2939delAG	p.840_850LDNQLQE	uc001gqx.2	P22
ELOVL2	54898	11103308	Missense	c.584G>T	p.Q141H	uc003mzp.2	P22
EPHB1	2047	136450026	Missense	c.2895C>A	p.A892E	uc003eqt.1	P22
GALNT6	11226	50045526	Missense	c.1090G>C	p.A257P	uc001ryl.1	P22
HAP1	9001	37141336	Frame_Shift_Ins	c.1015_1016insAA	p.A335fs	uc002hxm.1	P22
HVCN1	84329	109573510	Missense	c.703G>T	p.V180F	uc001trs.1	P22
ID2	3398	8739889	Missense	c.326_327AG>TT	p.E48V	uc002qza.1	P22
IQSEC1	9922	12952029	Missense	c.1538G>T	p.R510L	uc003bxt.1	P22
ITPR2	3709	26530428	Missense	c.6104C>A	p.P1896Q	uc001rhg.1	P22
KCNK2	3776	213326342	Missense	c.224C>T	p.P19S	uc001hkq.1	P22
KIF26B	55083	243597090	Missense	c.1237_1238GC>A	p.S266N	uc001ibf.1	P22
KRT19	3880	36933621	Missense	c.1245A>T	p.D368V	uc002hxd.2	P22
LAT	27040	28908406	Missense	c.990C>T	p.S213F	uc002dsd.1	P22
LIMK2	3985	29993012	Frame_Shift_Del	c.1376_1380delTT	p.L341fs	uc003akj.1	P22
MACF1	23499	39521533	Missense	c.1001G>T	p.G266W	uc009vvo.1	P22
MAGED2	10916	54854136	Frame_Shift_Del	c.789_807delCTC	p.T232fs	uc004dtk.1	P22
MCF2L2	23101	184408211	Missense	c.2681G>T	p.R864L	uc003fil.1	P22
MPI	4351	72969987	Missense	c.88C>T	p.A28V	uc002azc.1	P22
MURC	347273	102388017	Missense	c.648G>T	p.R186S	uc004bba.1	P22
PCDH8B	56128	140539046	Missense	c.1433C>T	p.A416V	uc003liu.1	P22
PITPNM2	57605	122039280	Frame_Shift_Del	c.2963_2963delC	p.L942fs	uc001uej.1	P22
PRKCD	5580	53190533	In_frame_Del	c.763_783delCCA	p.137_144AKFTPMTMN	uc003dgl.1	P22
PSMC5	5705	59262618	Missense	c.1031G>T	p.K330N	uc002jcb.1	P22
PTPRM	5797	7945388	Missense	c.1611C>A	p.L370I	uc010dkv.1	P22
SH3TC2	79628	148398234	Missense	c.970G>T	p.C273F	uc003ipu.1	P22
SPAG9	9043	46552921	Nonsense	c.174C>G	p.Y32*	uc002itzc.1	P22
UMOD	7369	20265034	Frame_Shift_Del	c.1324_1325delTG	p.C399fs	uc002dhb.1	P22
ZNF205	7755	3109866	Missense	c.1339A>C	p.T402P	uc002cub.1	P22
ZNF211	10520	62845282	Missense	c.1942G>T	p.C604F	uc002qps.1	P22
ZNF461	92283	41821838	Nonsense	c.1477G>T	p.E417*	uc002oem.1	P22
ZNF846	162993	9729483	Frame_Shift_Ins	c.1800_1801insGA	p.E423fs	uc002mmn.1	P22
ATM	472	107691965	Missense	c.6498A>G	p.H2038R	uc001pkb.1	P23
CPE	1363	166625050	Missense	c.1094C>T	p.P273S	uc003irg.2	P23
DDX19A	55308	68956002	Missense	c.574G>A	p.V149I	uc002eys.1	P23
DENND5A	23258	9148807	Missense	c.2255C>T	p.P667L	uc001mhl.1	P23
DHX57	90957	38903839	Missense	c.3190G>A	p.D1031N	uc002rrf.1	P23
ECT2	1894	173962997	Missense	c.878A>G	p.K286E	uc003fil.1	P23
ELAVL3	1995	11438604	Frame_Shift_Ins	c.427_428insG	p.G16fs	uc002mrny.1	P23
LAMP1	3916	113008873	Missense	c.415A>G	p.N45S	uc001vtm.1	P23
MED12	9968	70255426	Missense	c.296G>A	p.E33K	uc004ddy.1	P23
MPDZ	8777	13209623	Missense	c.1072C>T	p.R341C	uc010mhy.1	P23
SLIT2	9353	20134844	Missense	c.1588C>T	p.R462C	uc003gpr.1	P23
SMYD1	150572	88168522	Missense	c.343T>G	p.V114G	uc002ssr.1	P23
ANTXR2	118429	81125009	Frame_Shift_Ins	c.1599_1600insC	p.P358fs	uc003hlz.2	P24
BIRC6	57448	32554873	Missense	c.6943A>G	p.K2270R	uc010ezu.1	P24

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CAMLG	819	134102256	Missense	c.152T>G	p.V16G	uc003kzt.1	P24
CLSTN2	64084	141764403	Missense	c.2283G>A	p.R758H	uc003etn.1	P24
COL9A1	1297	71023190	Splice_Site_SNP	c.e21_splice_site		uc003pgf.2	P24
DMXL2	23312	49559613	Missense	c.6805C>A	p.Q2194K	uc002abf.1	P24
DNAH8	1769	38991084	Missense	c.10042C>A	p.L3148I	uc003ooe.1	P24
FAT3	120114	92171426	Missense	c.5616G>A	p.V1867I	uc001pdj.2	P24
GEMIN7	79760	50285598	Missense	c.537T>A	p.F129Y	uc002pap.1	P24
GPC6	10082	93478090	Missense	c.1433T>C	p.V273A	uc001vlt.1	P24
HNRNPU1	11100	46500507	Frame_Shift_Ins	c.2074_2075insGA	p.N595fs	uc002oqb.2	P24
HSPG2	3339	22087045	Missense	c.716A>T	p.R226W	uc009vqd.1	P24
KCTD7	154881	65741622	Missense	c.945C>T	p.P280S	uc003tve.1	P24
NAGLU	4669	37949472	Missense	c.2262A>C	p.N641T	uc002hzv.1	P24
NTF4	4909	54256756	Missense	c.452T>G	p.V104G	uc002pmf.2	P24
PCLO	27445	82423961	Missense	c.4533C>G	p.T1415R	uc003uhx.2	P24
PHF19	26147	122662211	Missense	c.1631A>C	p.T460P	uc004bks.1	P24
PLEKHG4B	153478	216536	Missense	c.2331G>T	p.V761L	uc003jak.2	P24
POLG	5428	87674485	Missense	c.968T>G	p.V229G	uc002bns.2	P24
RAPGEF2	9693	160493478	Missense	c.3884C>T	p.R1192W	uc003iqg.2	P24
RNF150	57484	142088318	Missense	c.1484A>G	p.N277S	uc003iio.1	P24
SH3PXD2B	285590	171813903	Missense	c.230T>G	p.V20G	uc003mrbr.1	P24
SLC9A2	6549	102691271	Frame_Shift_Del	c.2472_2472delG	p.R777fs	uc002tca.1	P24
ST6GAL2	84620	106826213	Missense	c.828A>C	p.N218T	uc002tdr.1	P24
TMEM88	92162	7699304	Frame_Shift_Del	c.196_199delITC	p.F63fs	uc002giy.1	P24
TNRC18	84629	5393918	Missense	c.2412T>G	p.V688G	uc003soi.2	P24
ZAP70	7535	97717445	Missense	c.1127C>T	p.P307L	uc002syd.1	P24
ZNF614	80110	57210966	Missense	c.2036G>A	p.G566D	uc002pyj.1	P24
BBS10	79738	75265672	Missense	c.308A>G	p.H75R	uc001syd.1	P25
CCDC85A	114800	56273448	Nonsense	c.1111C>G	p.Y203*	uc002rzrn.1	P25
CHCHD10	400916	22438440	Frame_Shift_Ins	c.363_364insC	p.Q95fs	uc002zzw.1	P25
CHL1	10752	418307	Splice_Site_SNP	c.e27_splice_site		uc003bot.1	P25
DLX6	1750	96473321	Splice_Site_SNP	c.e1_splice_site		uc003uom.1	P25
EFTUD2	9343	40284618	Missense	c.2840A>C	p.T937P	uc002ihn.1	P25
ITIH1	3697	52787996	Splice_Site_SNP	c.e4_splice_site		uc003dps.2	P25
LCT	3938	136277987	Missense	c.4657A>G	p.Y1549C	uc002tuu.1	P25
LILRB4	11006	59868382	Missense	c.1106T>A	p.F239I	uc010ers.1	P25
MGAT4C	25834	84897673	Missense	c.2212C>T	p.T321M	uc001tai.2	P25
MIB2	142678	1554448	Frame_Shift_Ins	c.2576_2577insA	p.E817fs	uc001agg.1	P25
MYD88	4615	38157645	Missense	c.794T>C	p.L265P	NM_002468	P25
RAB11FIP5	26056	73156170	Missense	c.2190G>C	p.G650A	uc002siu.2	P25
SDHAF2	54949	60962050	Splice_Site_SNP	c.e3_splice_site		uc001nrt.1	P25
SEH1L	81929	12938134	Missense	c.152G>C	p.R5P	uc002krq.1	P25
SLIT3	6586	168120518	Nonsense	c.1875C>T	p.R538*	uc010jjg.1	P25
ADAMTS10	81794	8556462	Missense	c.3017G>A	p.V915I	uc002mkj.1	P26
ARID4B	51742	233464430	Frame_Shift_Del	c.1084_1084delG	p.V196fs	uc001hwq.1	P26
CD36	948	80137255	Missense	c.1483T>G	p.F267V	uc003uhc.1	P26
CDK13	8621	40098992	Missense	c.3601A>G	p.M1107V	uc003thh.2	P26
CECR2	27443	16383308	Missense	c.1119T>A	p.S331R	uc010gqw.1	P26
CMYA5	202333	79122587	Missense	c.11800G>T	p.A3910S	uc003kgc.1	P26
FAM70A	55026	119329145	Frame_Shift_Del	c.275_275delC	p.P16fs	uc004eso.2	P26
KIAA1598	57698	118633781	Frame_Shift_Del	c.2399_2399delT	p.L634fs	uc001lcx.2	P26
MGAT4C	25834	84901503	Missense	c.1474A>T	p.D75V	uc001tai.2	P26
MYRIP	25924	40060572	Missense	c.273A>G	p.K3R	uc010hhw.1	P26
NPAS3	64067	32906141	Splice_Site_SNP	c.e4_splice_site		uc001wru.1	P26
PTPRN2	5799	157063530	Missense	c.2617C>T	p.R854W	uc003wno.1	P26
RAPGEF2	9693	160494480	Missense	c.4310G>A	p.G1334R	uc003iqg.2	P26
STT3A	3703	124979323	Missense	c.571G>A	p.R160Q	uc001qcd.1	P26
TMEM195	392636	15566366	Missense	c.352T>C	p.L61P	uc003stb.1	P26
ZNF677	342926	58432812	Missense	c.1165T>C	p.V327A	uc002qbf.1	P26
B3GAT3	26229	62145914	Missense	c.111G>T	p.G28C	uc001ntw.1	P27
COL24A1	255631	85973133	Missense	c.4927A>G	p.T1629A	uc001dlj.1	P27
DACH2	117154	85957731	Missense	c.1723A>G	p.T575A	uc004eew.1	P27
DST	667	56465026	Missense	c.1434G>C	p.E4608D	uc003pcz.2	P27
EGR2	1959	64243254	Missense	c.1488C>A	p.H384N	uc001jmi.1	P27
FOXP3	2309	108989631	Missense	c.843A>G	p.K176R	uc003psk.2	P27
IGSF1	3547	130246905	Missense	c.731G>A	p.C199Y	uc004ewd.1	P27
KIAA1632	57724	41733467	Missense	c.4809C>T	p.P1570L	uc002lbn.1	P27
LAS1L	81887	64664934	Missense	c.959C>T	p.A296V	uc004dwa.1	P27
MICAL1	64780	109874059	Missense	c.2808T>G	p.W852G	uc003ptj.1	P27
MYCBP2	23077	76735819	Missense	c.1515T>C	p.L475P	uc001vkf.1	P27
NOTCH1	4851	138510470	Frame_Shift_Del	c.7541_7542delCT	p.P2514fs	uc004chz.1	P27
PPM1A	5494	59819255	Missense	c.396C>A	p.S100R	uc001xew.2	P27
RAPGEF4	11069	173387259	Missense	c.491G>A	p.V102M	uc002uhv.2	P27
SCN2A	6326	165872675	Missense	c.748A>T	p.D153V	uc002udc.1	P27
SLC5A7	60482	107980751	Missense	c.750T>A	p.D158E	uc002tdv.1	P27

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TGS1	96764	56861981	Missense	c.1357A>G	p.I324V	uc003xsj.2	P27
UBP1	7342	33409058	Splice_Site_SNP	c.e15_splice_site		uc003cfq.2	P27
ZNF182	7569	47721598	Missense	c.1178A>G	p.I278V	uc004dir.1	P27
ABCB1	5243	87034082	Nonsense	c.903G>A	p.W162*	uc003uij.1	P28
ARHGAP21	57584	24948737	Frame_Shift_Ins	c.2526_2527insG	p.E697fs	uc001isb.1	P28
ARID4B	51742	233407765	Missense	c.3919G>A	p.V1141I	uc001hwq.1	P28
CARS	833	2979017	Missense	c.2473G>A	p.S800N	uc001lxf.1	P28
COL25A1	84570	109959922	Missense	c.1914G>A	p.V620I	uc010imd.1	P28
FZD5	7855	208340841	Missense	c.1278G>A	p.V290I	uc002vcj.1	P28
KYNU	8942	143428880	Missense	c.535T>A	p.N135K	uc002vtl.1	P28
PCDH1	5097	141229051	Missense	c.287C>A	p.A57D	uc003ilp.1	P28
SAMHD1	25939	34978851	Frame_Shift_Del	c.998_998delC	p.R290fs	uc002xgh.1	P28
VWF	7450	5998644	Missense	c.4451G>A	p.V1401I	uc001qnn.1	P28
ZFP36	7538	44590543	Missense	c.403T>A	p.S115R	uc002olh.1	P28
ANGPTL5	253935	101270859	Missense	c.1404T>C	p.F270L	uc001pgl.1	P29
CPNE3	8895	87632388	Splice_Site_SNP	c.e14_splice_site		uc003ydv.1	P29
FAT4	79633	126591624	Missense	c.10003T>G	p.Y3335D	uc003if2.2	P29
FIBP	9158	65408057	Missense	c.1111C>G	p.P339A	uc009yqu.1	P29
HHATL	57467	42709305	Missense	c.1604G>A	p.R486H	uc003clw.1	P29
MAPK1	5594	20457181	Missense	c.1187A>T	p.Y316F	uc002zvn.1	P29
MAPK1	5594	20457256	Missense	c.1112A>G	p.D291G	uc002zvn.1	P29
PPP2R3C	55012	34655686	Frame_Shift_Del	c.421_421delA	p.S23fs	uc001wss.1	P29
PRKCQ	5588	6593051	Missense	c.413A>T	p.K110I	uc001iji.1	P29
RHD	6007	25502530	Missense	c.990A>C	p.Y311S	uc009vro.1	P29
SCN3A	6328	165654908	Read-through	c.6493T>A	p.*2001K	uc002uxc.1	P29
ADAMTSL4	54507	148794535	Missense	c.1468G>A	p.G437D	uc009wlw.1	P30
AVIL	10677	56487479	Missense	c.1422C>T	p.R465W	uc001sqj.1	P30
CTSB	1508	11743148	Frame_Shift_Del	c.474_474delG	p.G60fs	uc003wul.1	P30
HERC2	8924	26151908	Nonsense	c.4760C>T	p.R1552*	uc001zbj.1	P30
MARK2	2011	63414276	Missense	c.369G>T	p.C16F	uc009yox.1	P30
NR4A1	3164	50734881	Missense	c.1659G>A	p.E222K	uc001rzq.1	P30
ZNF697	90874	119970191	Missense	c.170G>A	p.G19E	uc001ehy.1	P30
ZNF804A	91752	185510424	Missense	c.2650A>G	p.T686A	uc002uph.1	P30
ACTL7B	10880	110657143	Missense	c.889C>T	p.R297C	uc004bdi.1	P31
BTBD1	53339	81501564	Missense	c.985T>C	p.F261S	uc002bjn.1	P31
FANCA	2175	88385382	Missense	c.1331C>T	p.A430V	uc002fou.1	P31
GPAT2	150763	96054010	Missense	c.1784A>G	p.I521V	uc002svf.1	P31
GRIN2B	2904	13608660	Missense	c.2958C>T	p.R927W	uc001rbt.2	P31
MAP1A	4130	41601424	Missense	c.928G>A	p.R154H	uc001zrt.1	P31
MYD88	4615	38157645	Missense	c.794T>C	p.L265P	NM_002468	P31
OR4C12	283093	49959841	Missense	c.773G>A	p.R258H	uc001nhc.1	P31
PTRF	284119	37828403	Missense	c.398C>G	p.A80G	uc002hzo.1	P31
RAB4B	53916	45984445	Missense	c.1422G>A	p.E182K	uc002opf.1	P31
RUNX1	861	35086661	Frame_Shift_Del	c.1333_1333delT	p.S362fs	uc010gmu.1	P31
ZBTB6	10773	124713556	Missense	c.706C>G	p.S206C	uc004bnh.1	P31
CELF3	11189	149946321	Nonsense	c.1640C>A	p.Y282*	uc001eys.1	P32
CETN2	1069	151747056	Missense	c.551G>C	p.K168N	uc004fgq.1	P32
CSMD2	114784	33784266	Missense	c.9235C>A	p.Q3020K	uc001bxm.1	P32
EIF2B2	8892	74539853	Splice_Site_SNP	c.e2_splice_site		uc001xrc.1	P32
FAM117A	81558	45150022	Missense	c.843C>G	p.S254R	uc002ipk.1	P32
GPR87	53836	152495273	Missense	c.812C>T	p.R151W	uc003eyt.1	P32
IGSF3	3321	116944276	Missense	c.2604C>A	p.F633L	uc001eqq.1	P32
KIAA1109	84162	123380426	Missense	c.4184G>T	p.R1380L	uc003ieh.1	P32
MAP3K12	7786	52167053	Frame_Shift_Del	c.487_488delCT	p.P130fs	uc001sdn.1	P32
MUC2	4583	1082884	Missense	c.11816C>T	p.T3930M	uc001lsx.1	P32
PHKA1	5255	71717660	Missense	c.3890T>G	p.F1197V	uc004eax.2	P32
PNKP	11284	55062237	Missense	c.89G>C	p.E13Q	uc002pqph.1	P32
RBMB19	9904	112840590	Missense	c.2515C>G	p.R811G	uc009zwi.1	P32
SF3B1	23451	197975079	Missense	c.2146A>G	p.K700E	uc002uu.1	P32
SGCG	6445	22792811	Missense	c.738C>T	p.A205V	uc001uom.1	P32
SLCO1A2	6579	21336396	Missense	c.2300G>C	p.A527P	uc001res.1	P32
SPOP	8405	45051434	Missense	c.859G>A	p.D130N	uc002ipb.1	P32
TCHP	84260	108830838	Missense	c.917A>G	p.E255G	uc001tpn.1	P32
USP44	84101	94442635	Missense	c.1829T>C	p.M562T	uc001teg.1	P32
ZNF282	8427	148552323	Missense	c.1772A>C	p.N556T	uc003wfm.1	P32
ZNF664	144348	123063059	Missense	c.2245G>A	p.G139R	uc001ufz.1	P32
ZNF791	163049	12600115	Missense	c.934A>G	p.S258G	uc002kmu.2	P32
ACSL6	23305	131335216	Missense	c.1463C>T	p.R454W	uc003kvx.1	P33
ADAMTS10	81794	8574766	De_novo_Start_OutOfFrame	c.597C>T		uc002mkk.1	P33
ANKS6	203286	100570330	Missense	c.2017T>C	p.S666P	uc004ayu.1	P33
ANXA10	11199	169285876	Missense	c.230G>T	p.A29S	uc003irm.1	P33
BTNL9	153579	180412853	Missense	c.1001A>C	p.T262P	uc003mmt.1	P33
C11orf41	25758	33561604	Missense	c.3798A>C	p.N1225T	uc001mup.2	P33
CDH12	1010	22114407	Missense	c.594C>T	p.R46W	uc010iuc.1	P33

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
CDH5	1003	64981869	Missense	c.1000G>T	p.V282F	uc002eom.2	P33
COL11A1	1301	103119877	Frame_Shift_Ins	c.5357_5358insC	p.P1680fs	uc001dum.1	P33
DCLK1	9201	35246793	Missense	c.2388G>A	p.A726T	uc001uvf.1	P33
DTNA	1837	30599964	Missense	c.110A>G	p.T37A	uc010dmn.1	P33
EP300	2033	39877805	Missense	c.3235T>C	p.I947T	uc003azl.2	P33
FOXR1	283150	118356625	Missense	c.1052T>C	p.I276T	uc001pu.1	P33
HCFC1	3054	152878970	Missense	c.1522A>C	p.T332P	uc004fjp.1	P33
HOOK2	29911	12744473	Missense	c.581C>T	p.T137M	uc002muy.2	P33
KCNA10	3744	110862914	Missense	c.407A>G	p.K7E	uc001dzt.1	P33
KRT16	3868	37022385	Missense	c.221T>C	p.S28P	uc002hxg.2	P33
MAP1A	4130	41604668	Missense	c.4172A>C	p.E1235D	uc001zrt.1	P33
MAP3K15	389840	19308260	Missense	c.2550G>C	p.A305P	uc004czk.1	P33
MARK1	4139	218893202	Missense	c.2473G>A	p.V626I	uc009xdw.1	P33
NBEAL1	65065	203711189	Missense	c.739C>T	p.P223L	uc002uzt.2	P33
PDE3A	5139	20657852	Missense	c.1242G>T	p.C407F	uc001reh.1	P33
P14K2A	55361	99400858	Missense	c.663A>T	p.K202N	uc001kog.1	P33
PLIN1	5346	88014406	Missense	c.531C>T	p.A136V	uc002boh.1	P33
SNX7	51375	98923179	Missense	c.406G>C	p.E47Q	uc001drz.1	P33
TERT	7015	1347170	Missense	c.889A>C	p.R277S	uc003jcb.1	P33
TNNI1	7135	199647223	Missense	c.341G>A	p.R114H	uc009wzw.1	P33
TP53	7157	7517845	Missense	c.1012G>A	p.R273H	uc002qim.2	P33
WNK2	65268	95094762	Missense	c.5305C>T	p.R1769C	uc004ati.1	P33
C9orf86	55684	138854454	In_frame_Del	c.2418_2420delAG	p.K661del	uc004cj.1	P34
CCDC21	64793	26470129	Nonsense	c.1818G>T	p.E563*	uc001bls.1	P34
DCAF6	55827	166301494	Frame_Shift_Ins	c.2724_2725insC	p.G828fs	uc001gex.1	P34
DNMT3B	1789	30859284	Missense	c.2797C>T	p.R826C	uc002wyc.1	P34
DPY19L2	283417	62240621	Missense	c.2396G>A	p.A739T	uc001srp.1	P34
E2F3	1871	20595009	Missense	c.1322T>C	p.I332T	uc003nda.2	P34
EGR2	1959	64243338	Missense	c.1404G>A	p.E356K	uc001jmi.1	P34
GAB3	139716	153594097	Missense	c.718G>A	p.V224I	uc004fmk.1	P34
LGR5	8549	70264078	Missense	c.2069G>T	p.T674M	uc001swl.1	P34
LY9	4063	159050298	Missense	c.753C>T	p.P235S	uc001fwu.1	P34
MLXIP	22877	121184519	Frame_Shift_Ins	c.1244_1245insC	p.A339fs	uc001ubr.2	P34
MPHOSPH9	10198	122244914	Missense	c.1864T>A	p.L586Q	uc001uel.1	P34
NDUFA4	4697	10945050	Splice_Site_SNP	c.e2_splice_site		uc003srx.1	P34
PREX2	80243	69143820	Splice_Site_SNP	c.e12_splice_site		uc003xxv.1	P34
PSMC5	5705	59262461	Missense	c.954C>A	p.L305M	uc002jcb.1	P34
PURB	5814	44890554	Missense	c.932G>C	p.E307Q	uc003tme.1	P34
RBM39	9584	33776456	Missense	c.796A>T	p.D151V	uc002xeb.1	P34
RPS6KA6	27330	83259120	Splice_Site_SNP	c.e10_splice_site		uc004eej.1	P34
SPCS3	60559	177478252	Missense	c.127C>A	p.L111M	uc003jur.2	P34
SSTR4	6754	22965250	Missense	c.1194G>A	p.R377H	uc002wsr.2	P34
TET1	80312	70074514	Nonsense	c.2527C>G	p.Y674*	uc001jok.2	P34
TGDS	23483	94026580	Missense	c.1092T>A	p.I324K	uc001vlw.1	P34
TRIM4	89122	99354609	Missense	c.482C>A	p.H118N	uc003usd.1	P34
ACPT	93650	55989580	Missense	c.916A>C	p.T306P	uc002pta.1	P35
BRD7	29117	48920149	Missense	c.1039T>C	p.F340S	uc002ege.1	P35
CMY45	202333	79068135	Missense	c.7863A>T	p.K2597N	uc003kgc.1	P35
FBXW7	55294	153464851	Missense	c.1939G>A	p.G597E	uc003ims.1	P35
FBXW7	55294	153478425	Missense	c.989C>A	p.F280L	uc003ims.1	P35
HOOK2	29911	12735564	Missense	c.2027G>A	p.R619Q	uc002muy.2	P35
NCOR1	9611	15952824	Splice_Site_SNP	c.e19_splice_site		uc002gpo.1	P35
OPRM1	4988	154453921	Missense	c.1022A>G	p.K324R	uc003qpq.1	P35
PAG1	55824	82068007	Missense	c.722C>T	p.A4V	uc003ybz.1	P35
PGBD3	267004	50393887	Missense	c.2838G>C	p.G895A	uc009xoe.1	P35
RABGGTA	5875	23808717	Missense	c.873T>G	p.F151V	uc001wof.1	P35
RLBP1	6017	87559426	Missense	c.774T>C	p.S132P	uc002bnl.1	P35
RNF213	57674	75940090	Missense	c.4637A>T	p.I1472L	uc002jyh.1	P35
RYK	6259	135377265	Missense	c.1552G>A	p.C485Y	uc003eqc.1	P35
SORCS3	22986	106927913	Missense	c.2228C>G	p.H667Q	uc001kyi.1	P35
TCP11	6954	35211892	Missense	c.426A>G	p.Y82C	uc003okd.2	P35
VPS13A	23230	79171461	Splice_Site_SNP	c.e61_splice_site		uc004akr.1	P35
WDR72	256764	51784659	Missense	c.1208A>G	p.Q389R	uc002acj.2	P35
WSCD2	9671	107128162	Missense	c.1376A>C	p.N211T	uc001tms.1	P35
ZMYM3	9203	70386657	Nonsense	c.1282C>T	p.Q399*	uc004dzh.1	P35
ZNF648	127665	180293126	Missense	c.851A>C	p.T215P	uc001goz.1	P35
ZXDA	7789	57953019	Frame_Shift_Ins	c.773_774insC	p.P187fs	uc004dve.1	P35
CDK20	23552	89773928	Splice_Site_SNP	c.e7_splice_site		uc004apr.1	P36
CDT1	81620	8739944	Frame_Shift_Ins	c.901_902insC	p.A283fs	uc002flu.1	P36
CXADR	1525	17807360	Frame_Shift_Del	c.160_160delG	p.V14fs	uc002yki.1	P36
FGD1	2245	54513876	Missense	c.1258C>G	p.P175R	uc004dtg.1	P36
IGFBP6	3489	51777969	Frame_Shift_Del	c.267_267delG	p.E67fs	uc001sbu.1	P36
KLF8	11279	56308797	Missense	c.1402G>C	p.V181L	uc004dur.1	P36
NAV2	89797	19912305	Missense	c.2369A>G	p.T670A	uc009yhw.1	P36

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NBPF14	25832	146482257	Frame_Shift_Del	c.1015_1015delA	p.N333fs	uc001eqq.1	P36
RAB11FIP4	84440	26872303	Splice_Site_SNP	c.e5_splice_site		uc002hgn.1	P36
SIX4	51804	60250237	Missense	c.1987A>G	p.T663A	uc001xfc.2	P36
TRIP11	9321	91550648	Missense	c.1638C>A	p.L284I	uc001xzy.2	P36
AMPH	273	38469152	Missense	c.905A>G	p.H279R	uc003tgu.1	P37
DACH2	117154	85954861	Nonsense	c.1462C>T	p.R488*	uc004eev.1	P37
DDX3X	1654	41089660	Frame_Shift_Del	c.2085_2085delT	p.S410fs	uc004dfe.1	P37
GRID2	2895	94595938	Nonsense	c.1906C>T	p.R550*	uc003hsz.2	P37
IGSF22	283284	18695110	Missense	c.1177G>T	p.K329N	uc009yht.1	P37
MCAM	4162	118690941	Missense	c.241C>T	p.T71M	uc001pwf.1	P37
MICAL3	57553	16747051	Frame_Shift_Ins	c.1842_1843insC	p.R472fs	uc002znj.1	P37
MYT1L	23040	1822085	Missense	c.3750G>A	p.A975T	uc002qxe.1	P37
POLL	27343	103330004	Frame_Shift_Ins	c.2119_2120insAT	p.L451fs	uc001ktg.1	P37
PTPRB	5787	69251258	Missense	c.3229G>A	p.G1062E	uc001swc.2	P37
SCN2A	6326	165954314	Missense	c.6042C>T	p.R1918C	uc002udc.1	P37
SF3B1	23451	197975079	Missense	c.2146A>G	p.K700E	uc002uuue.1	P37
SUSD4	55061	221603326	In_frame_Del	c.697_699delGCA	p.21_22QQ>Q	uc001hnx.1	P37
ZC3H12B	340554	64638489	Missense	c.1162G>T	p.A385S	uc010nko.1	P37
NEU4	129807	242404444	Frame_Shift_Ins	c.577_578insC	p.V42fs	uc002wcn.1	P38
ZMYM3	9203	70389672	Frame_Shift_Del	c.246_246delC	p.S53fs	uc004dzh.1	P38
ABC B5	340273	20749137	Missense	c.3374T>C	p.V1046A	uc010kuh.1	P39
ACSS1	84532	24942689	Missense	c.2238G>A	p.A454T	uc002wub.1	P39
AKAP12	9590	151712279	Missense	c.1249G>A	p.E354K	uc003qoe.1	P39
ALDH1A1	216	74733730	Missense	c.393C>T	p.L114F	uc004ajd.1	P39
B3GALT1	8708	168434477	Missense	c.1033C>T	p.P228S	uc002udz.1	P39
BRD7	29117	489114113	Missense	c.1809C>T	p.L597F	uc002ege.1	P39
BSN	8927	49674601	Missense	c.10433G>C	p.S3440T	uc003cxe.2	P39
C2orf42	54980	70262594	Missense	c.356G>C	p.V10L	uc002sgh.1	P39
CCDC9	26093	52455748	Missense	c.420G>C	p.G92R	uc002pgph.1	P39
CDHR5	53841	609562	Missense	c.1310G>A	p.R402Q	uc001lqj.1	P39
CHD5	26038	6108495	Missense	c.4189T>G	p.D1363E	uc001amb.1	P39
CLCN1	1180	142758858	Missense	c.2732C>T	p.P882L	uc003wcr.1	P39
CPNE9	151835	9721438	Missense	c.286G>C	p.V39L	uc003bsd.1	P39
CR1	1378	205858200	Missense	c.7191G>A	p.D2351N	uc001hfx.1	P39
CSAD	51380	51852591	Missense	c.550C>T	p.R79C	uc001sbx.1	P39
DCHS2	54798	155383276	Missense	c.5675T>A	p.F1892Y	uc003inw.1	P39
DNMBP	23268	101638648	Missense	c.3301T>C	p.M1070T	uc001kqj.2	P39
DOLK	22845	130748773	Missense	c.1061C>T	p.R211C	uc004bw.1	P39
DST	667	56643470	Missense	c.1029G>A	p.G170E	uc003pcz.2	P39
EXOSC8	11340	36475070	Splice_Site_SNP	c.e4_splice_site		uc001uwa.1	P39
F5	2153	167796473	Missense	c.674A>G	p.N177D	uc001ggg.1	P39
GAB4	128954	15848875	Missense	c.769G>A	p.A221T	uc002zlw.1	P39
GALNT8	26290	4740568	Nonsense	c.1449C>T	p.Q453*	uc001qne.1	P39
GRIK5	2901	47238696	Missense	c.1356G>A	p.E441K	uc002osj.1	P39
HDAC4	9759	239701828	Missense	c.2426C>T	p.P545L	uc002vyk.2	P39
IFNA8	3445	21399358	Missense	c.213C>G	p.F61L	uc003zpc.1	P39
IGSF10	285313	152648274	Missense	c.2185C>T	p.R729C	uc003zeb.1	P39
JUB	84962	22513160	Missense	c.1803G>C	p.C476S	uc001whz.1	P39
KCNK13	56659	89720462	Missense	c.1031G>A	p.V197I	uc001xye.1	P39
KIF7	374654	87977985	Missense	c.1174G>A	p.R330H	uc002bof.1	P39
LIP1	149998	14476028	Missense	c.638T>G	p.F210V	uc002yjm.1	P39
LRRK1	79705	99385047	Missense	c.2783G>A	p.V822I	uc002bwr.1	P39
LUC7L	55692	196120	Missense	c.505G>A	p.E132K	uc002cgc.1	P39
MARCKS	4082	114288354	Missense	c.1300C>A	p.A302E	uc003pv.2	P39
MME	4311	156349110	Missense	c.1786G>T	p.K525N	uc010hvr.1	P39
PAX8	7849	113694145	Missense	c.1437T>G	p.L424W	uc002tjk.1	P39
PELI2	57161	55714897	Missense	c.455A>T	p.S57C	uc001xch.1	P39
PHRF1	57661	598503	In_frame_Del	c.3175_3186delG	p.TRSG1017del	uc001lqe.1	P39
POTE B	339010	19335787	Missense	c.881C>G	p.Q172E	uc001ytu.1	P39
PRMT6	55170	107401838	Missense	c.907C>G	p.N267K	uc001dvb.1	P39
PTPRU	10076	29503025	Missense	c.2688C>T	p.R860W	uc001bru.1	P39
RAPGEF1	2889	133491472	Missense	c.1522C>T	p.H455Y	uc004ccb.1	P39
RCL1	10171	4831330	Missense	c.941A>G	p.D228G	uc003zis.2	P39
RNF38	152006	36342814	Missense	c.1294C>T	p.T368I	uc003zzh.1	P39
RPL31	6160	100988954	Missense	c.422C>A	p.T112N	uc010fiu.1	P39
RYR3	6263	31865241	Missense	c.942G>A	p.E3119K	uc001zhi.1	P39
SERPINA12	145264	94034466	Missense	c.818G>A	p.A8T	uc001ydi.1	P39
SLC10A6	345274	87965636	Missense	c.880G>A	p.G294R	uc003hqd.1	P39
TBC1D8	11138	101037194	Missense	c.525G>A	p.E132K	uc010fiv.1	P39
TINAG	27283	54299621	Missense	c.718G>A	p.R191H	uc003pcj.1	P39
TP53	7157	7519251	Missense	c.598G>A	p.C135Y	uc002gim.2	P39
U2AF2	11338	60864312	Missense	c.1486T>A	p.M144K	uc002qlu.1	P39
UPP2	151531	158682585	Missense	c.534G>A	p.G115S	uc002lzo.1	P39
WDR73	84942	82987881	In_frame_Del	c.960_977delATG	p.DGTRSQ315del	uc002bkw.1	P39

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WNK4	65266	38201821	Missense	c.3607A>C	p.T1196P	uc002ibj.1	P39
WNK4	65266	38201824	Missense	c.3610T>C	p.S1197P	uc002ibj.1	P39
WWTR1	25937	150742960	Missense	c.639A>C	p.N208T	uc003exe.1	P39
ZNF556	80032	2828320	Missense	c.451C>T	p.R122C	uc002lwp.1	P39
ZNF777	27153	148783559	Missense	c.651A>G	p.D163G	uc003wfv.1	P39
ZNF793	390927	42720002	Missense	c.1044C>T	p.P201L	uc010efm.1	P39
ABLM2	84448	8072915	Missense	c.1327G>A	p.R395Q	uc003gko.2	P40
AMOTL2	51421	135563304	Nonsense	c.1599C>T	p.R439*	uc003eqg.1	P40
ASB18	401036	236787746	Frame_Shift_Ins	c.1098_1099insC	p.P366fs	uc010fyo.1	P40
BTBD3	22903	11848400	Missense	c.811C>T	p.A151V	uc002wnz.1	P40
CSMD1	64478	3251052	Missense	c.2563G>A	p.D725N	uc010lrh.1	P40
GRIK5	2901	47201867	Missense	c.2146G>A	p.S704N	uc002osj.1	P40
KIAA0226	9711	198913134	Splice_Site_SNP	c.e6_splice_site		uc003fyc.2	P40
KIAA1199	57214	79001412	Missense	c.2341G>A	p.G694E	uc002bfw.1	P40
KPNA5	3841	117129873	Splice_Site_SNP	c.e6_splice_site		uc003pxh.1	P40
OR5R1	219479	55941797	Missense	c.488C>T	p.T163I	uc001niu.1	P40
PTPRD	5789	8474233	Missense	c.4010C>T	p.T1100M	uc003zkk.1	P40
RGS2	5997	191045917	Splice_Site_SNP	c.e2_splice_site		uc001gsl.1	P40
RRP1B	23076	43935778	Missense	c.2164G>T	p.V684F	uc002zdk.1	P40
SF3B1	23451	197973694	Missense	c.2756A>G	p.Q903R	uc002uee.1	P40
TFCP2	7024	49789182	Missense	c.1165A>G	p.K236E	uc001rxw.1	P40
VWA3B	200403	98253575	Splice_Site_SNP	c.e22_splice_site		uc002syo.1	P40
XIRP2	129446	167823572	Missense	c.2458G>T	p.R790I	uc010fpn.1	P40
C6	729	41185830	Missense	c.2610C>A	p.S791Y	uc003jml.1	P41
CASP4	837	104327874	Missense	c.404C>T	p.H111Y	uc001pid.1	P41
CMKLR1	1240	107210118	Missense	c.1259G>A	p.R249H	uc001tmv.1	P41
DDR2	4921	160996394	Splice_Site_SNP	c.e9_splice_site		uc001gcf.1	P41
DRGX	644168	50244225	Missense	c.749G>A	p.G250D	uc001jhq.1	P41
FBN1	2200	46679698	Missense	c.700G>A	p.M124I	uc001zwx.1	P41
HERC3	8916	89808138	Missense	c.1682A>G	p.I506V	uc003hrw.1	P41
LANCL1	10314	211009349	Missense	c.990G>C	p.E296Q	uc002ved.1	P41
MCHR2	84539	100489018	Nonsense	c.999T>A	p.Y228*	uc003pqh.1	P41
NRXN1	9378	50700770	Missense	c.2691A>G	p.Y405C	uc002rxe.2	P41
NRXN2	9379	64175636	Missense	c.3024C>T	p.T862M	uc001oar.1	P41
PCDHAC2	56134	140369551	Missense	c.3109C>T	p.R957W	uc003ili.1	P41
PLEKHG3	26030	64278345	Missense	c.2626T>A	p.L786Q	uc001xho.1	P41
PMS2	5395	5992931	Missense	c.2078T>A	p.L664Q	uc003spl.1	P41
PTPRF	5792	43836122	Missense	c.2270G>A	p.V644M	uc001cjr.1	P41
RGS9	8787	60586832	Missense	c.335C>A	p.N75K	uc002ife.1	P41
RIPK1	8737	3058352	Missense	c.2028A>G	p.K599R	uc010jni.1	P41
SON	6651	33870612	Missense	c.6331G>C	p.A1405P	uc002ysd.2	P41
SPEG	10290	220056174	Frame_Shift_Ins	c.5745_5746insG	p.S1915fs	uc010fwg.1	P41
THUMPD2	80745	39850562	Missense	c.462T>G	p.I125R	uc002rru.1	P41
TP53	7157	7518293	Missense	c.907G>C	p.C238S	uc002gim.2	P41
ATF7IP	55729	14540430	Splice_Site_SNP	c.e14_splice_site		uc001rbw.1	P42
C3orf62	375341	49288927	Missense	c.530C>A	p.A128E	uc003cwn.1	P42
CALHM1	255022	105205258	Missense	c.929C>A	p.H264Q	uc001kxe.1	P42
CNOT1	23019	57150066	Missense	c.2437C>A	p.A715D	uc002env.1	P42
CREBFZ	58487	85052735	Missense	c.1087C>T	p.A278V	uc001pas.1	P42
CSNK1E	1454	37026883	Missense	c.823C>G	p.I119M	uc003avm.1	P42
ECT2L	345930	139243830	Missense	c.1812T>A	p.V570D	uc003qif.1	P42
EIF4ENIF1	56478	30181144	Missense	c.1421T>A	p.N419K	uc003akz.1	P42
ELN	2006	73112274	Missense	c.1646G>A	p.V519I	uc003tzw.1	P42
FBXW7	55294	153468834	Missense	c.1543G>A	p.R465H	uc003ims.1	P42
IFT140	9742	1513671	Missense	c.3349C>G	p.A1101G	uc002cm.1	P42
IL17RD	54756	57107155	Missense	c.1705G>A	p.G539D	uc003dil.1	P42
MACF1	23499	39662421	Missense	c.11735A>C	p.R3868S	uc009vv.1	P42
MPRIP	23164	16922048	Splice_Site_SNP	c.e3_splice_site		uc002gqv.1	P42
MUC5B	727897	1227452	Missense	c.12833A>C	p.T4259P	uc001ltb.2	P42
MYH11	4629	15725594	Missense	c.4418C>A	p.D1437E	uc002ddx.1	P42
NOVA1	4857	25987037	Missense	c.1810G>T	p.V498F	uc001wpy.1	P42
PCDHGB7	56099	140778868	Missense	c.1403G>A	p.V420I	uc003ln.1	P42
PDS5B	23047	32130391	Missense	c.486A>T	p.I110L	uc010abf.1	P42
PEG3	5178	62019991	Missense	c.1982T>C	p.F544S	uc002qnu.1	P42
PTPN21	11099	88015924	Missense	c.1935G>A	p.G535D	uc001xvw.2	P42
SIGLEC11	114132	55153421	Missense	c.1673C>T	p.L516F	uc002pre.1	P42
SRGAP1	57522	62807931	Missense	c.2620C>A	p.P855H	uc001sru.1	P42
TP53	7157	7517822	Missense	c.1035G>A	p.D281N	uc002gim.2	P42
TTN	7273	179350895	Missense	c.4485C>T	p.R1421W	uc002umr.1	P42
ANK2	287	114470861	Missense	c.3011A>T	p.S971C	uc003ibe.2	P43
ARL6IP1	23204	18716815	Missense	c.292A>T	p.M75L	uc002dfi.1	P43
BAZ2A	11176	55279361	Nonsense	c.5421C>T	p.Q1743*	uc001slq.1	P43
C20orf177	63939	57953517	Missense	c.1539T>A	p.V375E	uc002yba.1	P43
C2orf3	6936	75775063	Splice_Site_SNP	c.e6_splice_site		uc002sno.1	P43

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
C4orf7	260436	71134491	Read-through	c.341T>A	p.*86K	uc003hfd.1	P43
CCDC81	60494	85801189	Missense	c.1759T>A	p.I444K	uc001pbx.1	P43
CHD8	57680	20938613	Splice_Site_SNP	c.e23_splice_site		uc001was.1	P43
ENPP7	339221	75323693	Missense	c.676T>G	p.V219G	uc002jxa.1	P43
ESCO1	114799	17398202	Missense	c.2715T>A	p.L594Q	uc002kth.1	P43
EVPL	2125	71522748	Missense	c.2294T>C	p.V689A	uc002qjg.2	P43
LAMC2	3918	181466811	Missense	c.2121G>A	p.E603K	uc001gqa.2	P43
LCE1C	353133	151044529	Missense	c.101C>A	p.T17N	uc001fap.1	P43
LRP1	4035	55884745	Missense	c.11606C>T	p.R3714C	uc001snd.1	P43
MITF	4286	70097073	Missense	c.1663C>T	p.T516M	uc003dnz.1	P43
NEUROD1	4760	182251396	Missense	c.673C>T	p.A146V	uc002uof.1	P43
OR2K2	26248	113130506	Missense	c.29G>A	p.S10N	uc004bfd.1	P43
PCLO	27445	82346622	Missense	c.13910G>A	p.G4541R	uc003uhx.2	P43
PDE1A	5136	182759004	Missense	c.1574C>T	p.S475L	uc002uoq.1	P43
PLEKH2	130271	43791228	Frame_Shift_Del	c.2421_2425delTT	p.F771fs	uc002rte.2	P43
PLG	5340	161059381	Missense	c.916G>A	p.G285R	uc003gtm.2	P43
RIPK1	8737	3050831	Nonsense	c.1355C>T	p.Q375*	uc010jni.1	P43
SCML1	6322	17678161	Missense	c.855G>A	p.R177H	uc004cyb.1	P43
SEMA5B	54437	124123960	Missense	c.1601C>T	p.P433S	uc003efz.1	P43
SF3B1	23451	197975079	Missense	c.2146A>G	p.K700E	uc002uee.1	P43
SPATA19	219938	133217134	Splice_Site_SNP	c.e6_splice_site		uc001ggv.1	P43
TBCK	93627	107385230	Splice_Site_SNP	c.e11_splice_site		uc010ilv.1	P43
TPR	7175	184581453	Splice_Site_SNP	c.e24_splice_site		uc001grv.1	P43
TTC3	7267	37426856	Missense	c.1468A>T	p.S455C	uc002yvz.1	P43
VPS13C	54832	59955338	Splice_Site_SNP	c.e76_splice_site		uc002agz.1	P43
ZNF488	118738	47990876	Missense	c.500T>G	p.V113G	uc001jex.1	P43
C1D	10438	68127936	Missense	c.93A>G	p.E4G	uc002sea.2	P44
CSMD3	114788	113632265	Missense	c.4534G>T	p.A1459S	uc003ynu.1	P44
DUSP15	128853	29916435	Missense	c.238A>T	p.D54V	uc002wwu.1	P44
FASTK	10922	150405196	Missense	c.1450T>C	p.F451S	uc003wix.1	P44
HECW1	23072	43450545	Missense	c.1854G>A	p.E417K	uc003tid.1	P44
HSPG2	3339	22058700	Missense	c.5282A>C	p.T1748P	uc009vqd.1	P44
KIAA0649	9858	137519296	Frame_Shift_Del	c.3668_3668delG	p.W1040fs	uc004cfr.1	P44
LRP5L	91355	24087684	Splice_Site_Del	c.e2_splice_site		uc003abs.1	P44
MRPL39	54148	25881941	Missense	c.1015C>T	p.T334M	uc002ylh.1	P44
NOSTRIN	115677	169429609	Missense	c.2333C>A	p.H525Q	uc002uef.1	P44
NSD1	64324	176643469	Missense	c.6223A>G	p.T2029A	uc003mr.2	P44
PLCB1	23236	8613596	Frame_Shift_Del	c.883_883delG	p.G294fs	uc002wnb.1	P44
PLXNB1	5364	48426925	Missense	c.5522T>A	p.D1821E	uc003csv.1	P44
PRKD1	5587	29466373	In_frame_Ins	c.277_278insTCC	p.z32_33insSG	uc001wqh.1	P44
SCN8A	6334	50431460	Missense	c.2364C>A	p.T729N	uc001ryw.1	P44
SEMA6C	10500	149379079	Missense	c.530C>G	p.A77G	uc001ewv.1	P44
SLCO4A1	28231	60758516	Missense	c.470T>G	p.W89G	uc002ydb.1	P44
STOX1	219736	70322473	Missense	c.2945C>T	p.P982L	uc001joq.1	P44
ANKRD17	26057	74229410	Missense	c.1990C>A	p.H625N	uc003hgp.1	P45
EPHX3	79852	15199693	Missense	c.829G>A	p.R249H	uc002naq.1	P45
KCNT2	343450	194494121	Missense	c.3097A>G	p.K1013E	uc001gtd.1	P45
WBSR16	81554	74127316	Frame_Shift_Del	c.319_320delIGG	p.G65fs	uc003ubr.1	P45
ZNF496	84838	245558776	Missense	c.443G>A	p.D136N	uc009xgv.1	P45
ADAMTS1L	92949	18816273	Missense	c.138T>C	p.F10S	uc003znf.2	P46
DKK2	27123	108064750	Missense	c.1295G>A	p.R197H	uc003hyi.1	P46
DST	667	56444018	Missense	c.15795A>G	p.E5092G	uc003pcz.2	P46
IREB2	3658	76573361	Missense	c.2542A>T	p.M794L	uc002bdr.2	P46
ITGA2B	3674	39805263	Missense	c.3147G>A	p.E1039K	uc002igl.1	P46
JTB	10899	152216301	Missense	c.775T>G	p.W18G	uc001fds.1	P46
MYD88	4615	38157341	Missense	c.773C>T	p.P258L	NM_002468	P46
OR13C5	138799	106400824	Missense	c.692C>T	p.S231L	uc004bcd.1	P46
PATE2	399967	125153035	Missense	c.195C>T	p.S50F	uc001gcu.1	P46
PTPN3	5774	111193291	Missense	c.2077C>T	p.T685I	uc004bed.1	P46
TLK2	11011	58033179	Missense	c.2099A>C	p.I611L	uc010ddp.1	P46
ZNF182	7569	47720661	Missense	c.2115G>T	p.R590I	uc004dir.1	P46
ZNF253	56242	19863538	Missense	c.574C>T	p.T161I	uc002noj.1	P46
BICD2	23299	94521305	Missense	c.1499_1500TC>C	p.L481P	uc004asp.1	P47
ENPEP	2028	111683440	Missense	c.2234A>T	p.Y631F	uc003iab.2	P47
JMJD5	79831	27133712	Missense	c.853A>G	p.Q227R	uc002doh.1	P47
M6PR	4074	8987663	Splice_Site_SNP	c.e4_splice_site		uc001qvf.1	P47
MAPK1	5594	20490147	Missense	c.724G>A	p.D162N	uc002zvn.1	P47
SET	6418	130495886	Missense	c.921C>T	p.P227L	uc004bvt.2	P47
SLC6A5	9152	20624972	Missense	c.2259T>G	p.C662W	uc001mqd.1	P47
ZFP37	7539	114844902	Missense	c.1845G>A	p.C606Y	uc004bgm.1	P47
ZNF33B	7582	42409608	Nonsense	c.911C>T	p.Q226*	uc001jaf.1	P47
ANK2	287	114494351	Frame_Shift_Del	c.5228_5228delG	p.E1710fs	uc003ibe.2	P48
ATM	472	107627803	Frame_Shift_Del	c.2022_2022delT	p.L546fs	uc001pkb.1	P48
BCL9	607	145558227	Missense	c.2382G>A	p.G548S	uc001epq.1	P48

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
BRCA1	672	38499191	Missense	c.2083G>A	p.S628N	uc002ict.1	P48
CALR	811	12910993	Missense	c.205T>A	p.F46Y	uc002mvu.1	P48
INSM2	84684	35074753	Missense	c.1755G>T	p.G515V	uc001wth.1	P48
KATNA1	11104	149961169	Missense	c.944A>T	p.Y300F	uc003qmr.1	P48
OR1L1	26737	124464474	Missense	c.809G>T	p.R270I	uc004bms.1	P48
PC	5091	66374384	Frame_Shift_Del	c.2883_2883delA	p.P867fs	uc001oji.1	P48
PDE6C	5146	95408730	Missense	c.2257G>A	p.D707N	uc001kiu.2	P48
SCN10A	6336	38743465	Missense	c.2723A>G	p.N908S	uc003ciq.1	P48
SORCS3	22986	106897468	Missense	c.1633T>C	p.I469T	uc001kyi.1	P48
UBE3B	89910	108425285	Missense	c.1960G>A	p.D453N	uc001top.1	P48
VIPR2	7434	158522254	Missense	c.884C>T	p.A233V	uc003woh.1	P48
WHSC1L1	54904	38306379	Missense	c.1773A>C	p.T419P	uc003xli.1	P48
ZNF536	9745	35627300	Missense	c.1129G>C	p.G331R	uc002nsu.1	P48
ACSF3	197322	87694810	Missense	c.391C>A	p.R74S	uc010cig.1	P49
C3	718	6648740	Missense	c.2568C>A	p.P836T	uc002nmf.1	P49
CACNA1C	775	24843111	Frame_Shift_Ins	c.1469_1470insT	p.V386fs	uc009zdu.1	P49
CPSF1	29894	145596346	Missense	c.1174G>C	p.G242A	uc003zck.1	P49
ENO1	2023	8854633	Splice_Site_SNP	c.e3_splice_site		uc001apj.1	P49
GPS2	2874	7156874	Frame_Shift_Del	c.1172_1172delT	p.F303fs	uc002gfv.1	P49
LRRC41	10489	46523911	Missense	c.1249C>A	p.T402N	uc001cpn.1	P49
OPRD1	4985	29062032	Missense	c.1011C>T	p.R257W	uc001brf.1	P49
PBRM1	55193	52638056	Missense	c.1349A>G	p.D446G	uc003des.2	P49
PEAR1	375033	155140344	Missense	c.1187T>C	p.M1T	uc001fqj.1	P49
PPIL2	23759	20379227	Missense	c.1450T>G	p.I445S	uc002zvh.2	P49
SOD1	6647	31960703	Splice_Site_SNP	c.e3_splice_site		uc002ypa.1	P49
SPP2	6694	234624182	Missense	c.98A>G	p.M5V	uc002vvk.1	P49
SPTLC3	55304	13003045	Missense	c.796C>A	p.N169K	uc002wod.1	P49
TP53	7157	7519260	In_frame_Del	c.587_589delCAA	p.N131del	uc002gim.2	P49
C5orf4	10826	154180111	Missense	c.1169G>A	p.R60H	uc003lvr.1	P50
DDX46	9879	134180077	Missense	c.2663C>G	p.A832G	uc003kzw.1	P50
FAM83C	128876	33338434	Missense	c.1680G>A	p.G521E	uc002xca.1	P50
HMP19	51617	173467064	Missense	c.611G>C	p.A156P	uc003mcx.1	P50
ILF3	3609	10659203	Missense	c.1600G>C	p.R50P	uc002mpq.1	P50
ITGB8	3696	20410824	Missense	c.2441A>G	p.E579G	uc003suu.1	P50
LRRC32	2615	76049852	Missense	c.676C>G	p.L145V	uc001oxq.2	P50
ME1	150365	40510635	Missense	c.3272G>C	p.A1083P	uc003baz.1	P50
MPL	4352	43591008	Missense	c.1931T>C	p.L629P	uc001ciw.1	P50
MUC2	4583	1083364	Missense	c.12296C>G	p.T4090S	uc001lsx.1	P50
MYBPC2	4606	55655181	Missense	c.2915C>G	p.A955G	uc002psf.2	P50
OR1Q01	219960	57752024	Missense	c.900G>T	p.K300N	uc001nmp.1	P50
PTPRD	5789	8426680	Missense	c.4709G>A	p.S133N	uc003zkk.1	P50
SIN3B	23309	16850080	Missense	c.3151T>G	p.V1046G	uc002ney.1	P50
SPINK7	84651	147673154	Splice_Site_Del	c.e2_splice_site		uc003lpd.1	P50
STIM1	6786	3833690	Splice_Site_Del	c.e1_splice_site		uc001lyv.1	P50
VSIG4	11326	65159001	Missense	c.1156C>G	p.C343W	uc004dwh.2	P50
BCL9	607	145558977	Missense	c.3132C>T	p.R798W	uc001epq.1	P51
CCDC147	159686	106156524	Missense	c.2373A>G	p.K747E	uc001kyh.1	P51
CDH10	1008	24629205	Missense	c.484G>A	p.R51H	uc003jgr.1	P51
CHKB	1120	49364755	Missense	c.1263A>G	p.Q360R	uc003bms.1	P51
CLDN5	7122	17891684	Missense	c.565T>G	p.V32G	uc002zpu.1	P51
DAO	1610	107801604	In_frame_Del	c.1074_1085delICC	p.LRGA255del	uc001tnp.1	P51
DDX11	1663	31129252	Missense	c.814A>G	p.E188G	uc001rjt.1	P51
DIP2A	23181	46743126	Missense	c.762G>C	p.A203P	uc002zjo.1	P51
HAPLN1	1404	82973138	Missense	c.1069G>A	p.R333H	uc003kim.1	P51
HEATR5B	54497	37069379	Missense	c.5921G>C	p.R1942T	uc002rpp.1	P51
HEPACAM	220296	124300050	Missense	c.617C>G	p.L71V	uc001qbk.1	P51
HSPG2	3339	22077280	Missense	c.2714G>C	p.A892P	uc009qvq.1	P51
KCNK10	54207	87799346	Missense	c.560C>T	p.R119W	uc001xwn.1	P51
KIAA0247	9766	69195053	De_novo_Start_OutOfFrame	c.304C>T		uc001xlk.1	P51
ME1	4199	84004180	Missense	c.1107T>G	p.V334G	uc003pjy.1	P51
PCDH15	65217	55257313	Missense	c.4608C>T	p.R1405C	uc001iju.1	P51
PDE3A	5139	20413828	Missense	c.365C>A	p.P115T	uc001reh.1	P51
PLXNA4	91584	131538055	Missense	c.2705T>G	p.C826G	uc003vra.2	P51
PTCD2	79810	71651988	Missense	c.33C>G	p.A8G	uc003kcb.1	P51
PTPRB	5787	69267212	Missense	c.2197C>T	p.S718F	uc001swc.2	P51
RBAK	57786	5070610	Missense	c.1321A>G	p.T333A	uc010kss.1	P51
RPS2	6187	19526110	Missense	c.786A>G	p.R200G	uc002cnn.2	P51
SF3B1	23451	197974856	Missense	c.2273G>A	p.G742D	uc002uu.1	P51
STC2	8614	172677727	Missense	c.1948C>T	p.S213L	uc003mco.1	P51
UBASH3B	84959	122165102	Missense	c.1216A>G	p.M286V	uc001ypy.2	P51
ZC3H18	124245	87171123	Missense	c.238G>T	p.D31Y	uc002fkj.1	P51
ABT1	29777	26706674	Missense	c.672G>A	p.R214H	uc003nii.1	P51
ANO2	57101	5542803	Missense	c.2995G>C	p.A975P	uc001qnm.1	P52
C9orf150	286343	12811410	Missense	c.1041G>A	p.R113K	uc003zkw.1	P52

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
CECR2	27443	16411744	Missense	c.4366G>A	p.A1414T	uc010gqw.1	P52
ERCC4	2072	13933620	Missense	c.1088A>G	p.K360R	uc002dce.2	P52
FAM160A2	84067	6189665	Missense	c.2967C>T	p.R870W	uc001mck.2	P52
GIGYF2	26058	233420510	Missense	c.3999G>C	p.Q1244H	uc002vtj.2	P52
GNB1	2782	1727802	Missense	c.571T>C	p.I80T	uc001aif.1	P52
HIST1H1E	3008	26264811	In_Frame_Del	c.274_279delGAC	p.DV72del	uc003ngq.1	P52
KIAA1045	23349	34962515	Missense	c.762G>C	p.S184T	uc003zvr.1	P52
LPHN1	22859	14131965	Missense	c.2070C>T	p.R592W	uc002mvg.1	P52
LPHN2	23266	82225604	Nonsense	c.3717T>G	p.Y1167*	uc001div.1	P52
MAGEB4	4115	30170708	Missense	c.619G>C	p.V179L	uc004dcdb.1	P52
MON1A	84315	49924022	Missense	c.783T>C	p.M185T	uc003cxz.1	P52
MTUS1	57509	17645436	Missense	c.2678T>G	p.D748E	uc003wxv.1	P52
NLGN3	54413	70300751	Missense	c.1005G>A	p.G234D	uc004dzb.1	P52
NLRP3	114548	245653155	Missense	c.405G>T	p.G95V	uc001icr.1	P52
OBSL1	23363	220136503	Missense	c.2555G>T	p.R833L	uc010fwk.1	P52
OLFML2A	169611	126612507	Missense	c.2067G>A	p.V652I	uc004bov.1	P52
RFTN1	23180	16394263	Missense	c.1074C>A	p.N264K	uc003cay.1	P52
SI	6476	166247361	Missense	c.1911A>C	p.T617P	uc003fei.1	P52
SLC24A3	57419	19612921	Missense	c.1200A>C	p.T335P	uc002wr1.1	P52
TADA2B	93624	7106703	Missense	c.435C>G	p.A95G	uc003gjw.2	P52
TANC1	85461	159662530	Missense	c.471C>T	p.S66F	uc002uag.1	P52
TAS1R1	80835	6562125	Missense	c.2420A>G	p.Y807C	uc001ant.1	P52
TLR8	51311	12848204	Missense	c.1329G>T	p.R393I	uc004cvd.1	P52
TMEM45A	55076	101758306	Missense	c.450A>G	p.E84G	uc003dua.1	P52
VGLL1	51442	135458735	Missense	c.706C>A	p.A179D	uc004ezy.1	P52
ZFP64	55734	50134673	Missense	c.2117G>A	p.V590I	uc002xwk.1	P52
ZHX1	11244	124336389	Frame_Shift_Del	c.1409_1409delC	p.Q327fs	uc003yqe.1	P52
AK1	203	129674895		c.255C>A	p.Y34*	uc004bsm.2	P53
ATP6V1A	523	114991359	Missense	c.1036G>A	p.E324K	uc003ea0.1	P53
CAMK1G	57172	207852798	Missense	c.1488C>G	p.S462R	uc001hhd.1	P53
CUL7	9820	43114581	Missense	c.4720C>A	p.L1473M	uc003otq.1	P53
DCAF8	50717	158476198	Missense	c.809C>G	p.S212R	uc001fvn.1	P53
DLG1	1739	198279777	Missense	c.1422C>G	p.C386W	uc003fxm.2	P53
FAM71E1	112703	55662825	Missense	c.971A>C	p.T205P	uc002psh.1	P53
GAK	2580	874327	Nonsense	c.1273C>A	p.Y358*	uc003gbm.2	P53
GTF2H1	2965	18336153	Missense	c.1499C>A	p.Q447K	uc001moh.1	P53
NEK10	152110	27328660	Missense	c.776G>T	p.V168L	uc003cdt.1	P53
SHB	6461	37964819	Missense	c.1422A>G	p.E285G	uc004aaax.1	P53
SNX1	6642	62213964	Missense	c.1306C>A	p.Q424K	uc002amv.1	P53
TLN2	83660	60898861	Missense	c.6865G>A	p.E2289K	uc002alb.2	P53
TMCO4	255104	19979805	Missense	c.276C>G	p.P12A	uc001bcn.1	P53
TTF1	7270	134257325	Missense	c.1998G>T	p.S649I	uc004cbl.1	P53
UBR4	23352	19288057	Missense	c.14217T>G	p.V4738G	uc001bbi.1	P53
ULK4	54986	41263441	Missense	c.4012C>A	p.Q1271K	uc003ckv.2	P53
WHSC1L1	54904	38308135	Missense	c.1554C>A	p.Q346K	uc003xli.1	P53
ZNF628	89887	60686239	Missense	c.2420A>C	p.T619P	uc002qld.2	P53
ALG1	56052	5073760	Splice_Site_SNP	c.e12_splice_site	uc002cyn.1	P54	
ANK3	288	61505733		c.5104T>C	p.S1638P	uc001jk.1	P54
ANKRD30A	91074	37461178	Missense	c.446G>A	p.S116N	uc001iza.1	P54
ANO6	196527	44068315	Missense	c.1472G>T	p.A424S	uc001roo.1	P54
ASPM	259266	195382133	Missense	c.155C>G	p.P20A	uc001gtu.1	P54
ATF2	1386	175690980	Missense	c.693C>T	p.T144I	uc002uij.1	P54
BEND2	139105	18131898	Missense	c.705C>G	p.P184R	uc004cy.2	P54
C4orf39	152756	166097930	Missense	c.381C>G	p.S102R	uc003iqx.1	P54
C9orf152	401546	112009610	Missense	c.625A>G	p.E3G	uc004beo.2	P54
CD163L1	283316	7440251	Missense	c.1783T>G	p.V586G	uc001qsy.1	P54
CDCA2	157313	25381826	Missense	c.1194A>G	p.T239A	uc003xep.1	P54
COL1A2	1278	93892413	Missense	c.3193C>T	p.P908S	uc003ung.1	P54
CYP4V2	285440	187359341	Missense	c.1142G>A	p.E280K	uc003iyw.2	P54
DBN1	1627	176817699	Missense	c.2030C>G	p.T583S	uc003mgx.2	P54
FAM129B	64855	129327247	Missense	c.534T>G	p.V111G	uc004brh.1	P54
FAM83B	222584	54913393	Missense	c.1781A>C	p.E555D	uc003pck.1	P54
GDAP2	54834	118264329	Missense	c.377T>C	p.W59R	uc001ehf.1	P54
GPATCH8	23131	39832053	Missense	c.2982G>A	p.R973K	uc002igw.1	P54
GPR135	64582	59000331	Missense	c.1482A>G	p.D456G	uc010apj.1	P54
HPSE2	60495	100364751	Missense	c.1280T>C	p.L407S	uc001kpn.1	P54
IQSEC2	23096	53296786	Missense	c.1898G>T	p.G566V	uc004dsd.1	P54
IRS4	8471	107864076	Missense	c.2233C>A	p.P719T	uc004eoc.1	P54
JPH4	84502	23109985	Missense	c.2572G>C	p.A599P	uc001wkr.1	P54
KIAA1467	57613	13100124	Missense	c.433G>C	p.S137T	uc001rb1.1	P54
MIA3	375056	220867582	Missense	c.406C>T	p.H133Y	uc001hnl.1	P54
NRG1	3084	32740945	Missense	c.1913G>T	p.S474I	uc003xiu.1	P54
ORMDL2	29095	54499049	Splice_Site_SNP	c.e2_splice_site	uc001shw.1	P54	
OTOF	9381	26555972		c.2093C>T	p.R656W	uc002rhk.1	P54

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
PLOD1	5351	11937535	Nonsense	c.732T>A	p.L214*	uc001atm.1	P54
WDR78	79819	67071951	Missense	c.1974G>C	p.A640P	uc001dcx.1	P54
ZNF155	7711	49187582	Nonsense	c.263G>T	p.E20*	uc002oxy.1	P54
8-Sep	23176	132122134	Missense	c.1538T>G	p.S434A	uc003kxx.2	P55
ACBD3	64746	224413665	Missense	c.793C>G	p.A249G	uc001hpy.1	P55
ADCY5	111	124504619	Missense	c.2697C>G	p.S899R	uc003egh.1	P55
AOC3	8639	38260200	Missense	c.1970C>T	p.R604C	uc002lrv.1	P55
ARHGEF1	9138	47091286	Missense	c.937G>A	p.R283Q	uc002osb.1	P55
ARHGEF2	9181	154194296	Missense	c.1934A>G	p.D560G	uc001fmu.1	P55
BCOR	54880	39806972	Nonsense	c.4436G>T	p.E1382*	uc004den.2	P55
C17orf64	124773	55861506	Missense	c.512T>G	p.V34G	uc002lyq.1	P55
C6orf27	80737	318448067	Missense	c.1709G>C	p.A491P	uc003nxb.2	P55
CD6	923	60533671	Missense	c.997T>G	p.V278G	uc001nqq.1	P55
CHD2	1106	91300817	Missense	c.2509C>T	p.T645M	uc002bsp.1	P55
DUOX2	50506	431913111	Missense	c.663A>G	p.R154G	uc010bea.1	P55
EGFL8	80864	32243180	Missense	c.782A>G	p.E226G	uc003oac.1	P55
EGFR	1956	55191052	Missense	c.1171C>G	p.R309G	uc003tqk.1	P55
EPHB6	2051	142274181	Missense	c.2234A>C	p.T468P	uc003wbq.1	P55
FAM120A	23196	95329296	Missense	c.1482G>A	p.G486E	uc004atw.1	P55
FCGBP	8857	45057933	Missense	c.14149C>A	p.T4714N	uc002omp.2	P55
FRMD7	90167	131055783	Missense	c.528G>A	p.C117Y	uc004ewn.1	P55
FRYL	285527	48206847	Missense	c.8985G>A	p.E2794K	uc003gyh.1	P55
GJB1	2705	70360607	Nonsense	c.420G>T	p.E109*	uc004dzf.2	P55
GLB1	2720	33074745	Missense	c.690C>G	p.S191R	uc003cfi.1	P55
GRIN2C	2905	70354510	Missense	c.2302A>C	p.T716P	uc002jl.1	P55
GUCY1A3	2982	156870986	Splice_Site_Del	c.e11_splice_site		uc003iov.1	P55
HAS3	3038	67705822	Missense	c.1038G>C	p.A272P	uc010cfh.1	P55
HCN3	57657	153521711	Missense	c.1229C>G	p.S407R	uc001fjz.1	P55
HOXA11	3207	27190885	Missense	c.476T>C	p.V135A	uc003syx.1	P55
KRAS	3845	25289548	Missense	c.219G>A	p.G13D	uc001grp.1	P55
LRBA	987	151946990	Nonsense	c.5875G>T	p.E1801*	uc010ipj.1	P55
PODNL1	79883	13904594	Missense	c.1737T>G	p.V488G	uc002mxr.1	P55
REPIN1	29803	149700156	Missense	c.1257G>C	p.G355A	uc010lpr.1	P55
SFT2D1	113402	166663046	Missense	c.202C>T	p.P58S	uc003qux.1	P55
SLC24A6	80024	112228736	Missense	c.1649G>C	p.R480P	uc001tvc.1	P55
STOML2	30968	35092804	Missense	c.125C>T	p.S21F	uc003zwi.1	P55
UNC5D	137970	35660758	Missense	c.1050G>A	p.R241K	uc003xjr.1	P55
C16orf93	90835	30676404	Missense	c.1416T>G	p.V362G	uc002dzn.1	P56
EPHA7	2045	94013300	Missense	c.2870T>G	p.I886R	uc003poe.1	P56
EXOC4	60412	133230962	Missense	c.1840A>G	p.D602G	uc003vrk.1	P56
PKD1L1	168507	47863826	Missense	c.4492C>T	p.H1498Y	uc003tny.1	P56
RBM28	55131	127767030	Missense	c.285A>T	p.D57V	uc003vmp.2	P56
SPEF2	79925	35828295	Missense	c.4655C>T	p.T1515I	uc003jjo.1	P56
SYCP1	6847	115254564	Nonsense	c.1553T>G	p.Y448*	uc001efr.1	P56
SYNE1	23345	152597570	Missense	c.21057G>A	p.E6819K	uc010kiw.1	P56
TMEM67	91147	94869261	Missense	c.1476C>T	p.P466S	uc003ygd.2	P56
TRAK2	66008	201957085	Missense	c.2509C>T	p.T688I	uc002uyb.2	P56
ACTB	60	5535517	Missense	c.200G>C	p.G55A	uc003sos.2	P57
C5	727	122784822	Missense	c.3352G>A	p.V1108I	uc004bkv.1	P57
C9orf98	158067	134688446	Missense	c.1412G>A	p.A286T	uc004cbu.1	P57
DTX2	113878	75950336	Missense	c.1400T>C	p.S282P	uc003uff.2	P57
FAM47A	158724	34059857	Missense	c.493G>T	p.D154Y	uc004ddg.1	P57
GTPBP8	29083	114192654	Missense	c.165C>G	p.P40A	uc003dzn.1	P57
MTERFD3	80298	105895678	Missense	c.2764G>T	p.Q315H	uc001tme.1	P57
NAA40	79829	63478517	Missense	c.831G>C	p.C235S	uc009yo.1	P57
ODF2L	57489	86625253	Missense	c.393T>G	p.C16G	uc001dln.1	P57
PKD1	5310	2100723	Missense	c.4655G>C	p.Q1482H	uc002cos.1	P57
PLEKHG3	26030	64268907	Missense	c.1491G>T	p.A408S	uc001xho.1	P57
PRKG2	5593	82293862	Missense	c.964G>T	p.G317V	uc003hmh.1	P57
PTAFR	5724	28349788	Missense	c.459T>G	p.I111S	uc001bpl.1	P57
RPGR	6103	38030527	In_frame_Del	c.2835_2837delG	p.889_890EE>E	uc004ded.1	P57
SMC1A	8243	53439984	Missense	c.2819C>A	p.T917N	uc004dsg.1	P57
SON	6651	33849541	Missense	c.6183G>C	p.R2045T	uc002ye.1	P57
TFR2	7036	100066571	Missense	c.1188A>G	p.S383G	uc003uvv.1	P57
TP63	8626	191069813	Missense	c.1225G>A	p.R379H	uc003fr.2	P57
TTC7B	145567	90225656	Missense	c.1053C>T	p.R311C	uc001xyp.1	P57
XIRP2	129446	167823940	Missense	c.2826G>A	p.V913I	uc010fpn.1	P57
XKR5	389610	6666955	Missense	c.675T>G	p.V218G	uc003wqp.1	P57
ATP8A2	51761	25015445	Missense	c.938C>T	p.P266S	uc001uqk.1	P58
CDC14B	8555	98324609	Missense	c.1795C>G	p.T448R	uc004awj.1	P58
CELF4	56853	33109144	Missense	c.905G>A	p.R170H	uc002lae.2	P58
CYB5R4	51167	84687597	Missense	c.774A>T	p.L214F	uc003pkf.1	P58
DAB2	1601	39411864	Missense	c.2770C>A	p.Q747K	uc003jik.2	P58
DNER	92737	229980218	Missense	c.1844C>T	p.T566M	uc002pv.1	P58

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
GATA5	140628	60473859	Missense	c.1032G>C	p.A324P	uc002ycx.1	P58
GCNT4	51301	74361401	Missense	c.1079G>C	p.C73S	uc003kdn.1	P58
IMPG1	3617	76771906	Missense	c.1083C>T	p.P318L	uc003pik.1	P58
MLL5	55904	104539509	Missense	c.4604C>T	p.P1357L	uc003vcm.1	P58
MNS1	55329	54510964	Missense	c.1459T>G	p.L432V	uc002adr.1	P58
MYC	4609	128819862	Missense	c.741A>G	p.T73A	uc003ysi.1	P58
MYO9A	4649	69957617	Missense	c.6222G>A	p.G1917R	uc002atl.2	P58
PREPL	9581	44413214	Missense	c.1276C>T	p.P414L	uc002ruf.1	P58
SF3B1	23451	197974958	Missense	c.2267G>A	p.G740E	uc002uu.1	P58
SREBF1	6720	17663713	Missense	c.859C>T	p.S222F	uc002grt.1	P58
SRRM3	222183	75732067	Missense	c.932G>C	p.K241N	uc003uer.2	P58
8-Sep	23176	132122134	Missense	c.1538T>G	p.S434A	uc003kxu.2	P59
ABCC9	10060	21980741	Missense	c.155G>T	p.L45F	uc001frh.1	P59
ACACB	32	108174243	Missense	c.5919C>G	p.R1934G	uc001tob.1	P59
ADH1C	126	100479799	Missense	c.1146A>G	p.E354G	uc003huu.1	P59
ALS2	57679	202278388	Missense	c.4821A>C	p.K1541T	uc002uyo.1	P59
AMBN	258	71497342	Missense	c.197G>A	p.S41N	uc003hfl.1	P59
ARAP3	64411	141021488	Missense	c.3144C>G	p.C1022W	uc003llm.1	P59
ASPM	259266	195364414	Missense	c.2862G>A	p.S922N	uc001gtu.1	P59
ATXN7L3	56970	39630128	Missense	c.441A>C	p.N117T	uc002ifz.1	P59
BAT2L1	84726	133342991	Missense	c.4501C>G	p.C1482W	uc004can.2	P59
C10orf2	56652	102738034	Missense	c.733G>C	p.G26A	uc001ksf.1	P59
C16orf7	9605	88303277	Missense	c.1581A>C	p.T486P	uc002fom.1	P59
C16orf79	283870	2199695	Missense	c.629G>T	p.W151L	uc010bsh.1	P59
CADM2	253559	86093417	Missense	c.879T>A	p.N293K	uc003dqj.1	P59
CADM2	253559	86197508	Missense	c.1133T>G	p.V378G	uc003dqj.1	P59
CCDC27	148870	3670243	Missense	c.1519C>A	p.Q479K	uc001akv.1	P59
CDHR5	53841	608063	Missense	c.2114C>G	p.A670G	uc001lqj.1	P59
CDK17	5128	95241998	Missense	c.631C>G	p.P48A	uc001tep.1	P59
COBL	23242	51255118	Missense	c.244G>C	p.R20P	uc003ptr.2	P59
COL5A1	1289	136806558	Nonsense	c.2746C>A	p.Y788*	uc004cfe.1	P59
CSRP2BP	57325	18071545	Missense	c.863C>A	p.Q81K	uc002wj.1	P59
DAZAP1	26528	1385835	Missense	c.1337G>C	p.G383A	uc002lsn.1	P59
DSCAM	1826	40387535	Missense	c.4285T>G	p.V1278G	uc002yyq.1	P59
ERBB2IP	55914	65410018	Nonsense	c.4209C>A	p.Y1384*	uc010iwx.1	P59
FAMB48	157638	127638104	Missense	c.997G>C	p.R238P	uc003yrz.1	P59
FGF3	2248	69334469	Missense	c.996A>C	p.T169P	uc001oph.1	P59
FZD5	7855	208341571	Nonsense	c.548C>A	p.Y46*	uc002vcj.1	P59
GRPEL1	80273	7113630	Missense	c.555C>T	p.P172S	uc003gjy.1	P59
HEATR7B2	133558	41054271	Missense	c.3182G>T	p.V898F	uc003jmj.2	P59
HIST1H1T	3010	26216200	Missense	c.144G>C	p.S34T	uc003ngj.1	P59
HMG20A	10363	75557872	Missense	c.1073T>G	p.V291G	uc002bcr.1	P59
INSL3	3640	17788847	Missense	c.312A>G	p.R103G	uc010ebf.1	P59
ITGA10	8515	144239962	Missense	c.478C>G	p.S134R	uc001eo.1	P59
ITGAX	3687	31298601	Missense	c.2958A>C	p.D964A	uc002ebt.2	P59
KCNK15	60598	42808189	Missense	c.288G>C	p.G75A	uc002xmr.1	P59
KIAA1267	284058	41472921	Missense	c.2282A>C	p.T733P	uc002ikb.1	P59
LANCL3	347404	37403650	Missense	c.1016G>T	p.L238F	uc004ddp.1	P59
LMTK2	22853	97661455	Missense	c.4035T>C	p.S1248P	uc003upd.1	P59
MAPK7	5598	19224729	Missense	c.968G>C	p.R205P	uc002gvn.1	P59
MLPH	79083	238125802	Missense	c.1986G>C	p.A587P	uc002vvt.1	P59
MYH4	4622	10308535	Missense	c.738A>C	p.E209D	uc002gmn.1	P59
MYOM1	8736	3119302	Missense	c.3056A>C	p.T908P	uc002klp.1	P59
NANOS3	342977	13849199	Missense	c.250T>G	p.L46R	uc002mxj.2	P59
NUP160	23279	47813840	Missense	c.1125C>T	p.A347V	uc001ngm.1	P59
OBSCN	84033	226529063	Missense	c.5895G>C	p.A1951P	uc009xez.1	P59
PCDHGB7	56099	140777657	Missense	c.1927T>G	p.V16G	uc003ln.1	P59
PITPNM3	83394	6316797	Missense	c.1484G>C	p.E445Q	uc002gdd.2	P59
PPP1R12C	54776	60315715	Missense	c.519A>C	p.D168A	uc002qix.1	P59
PPP1R9A	55607	94741195	Missense	c.3455T>G	p.V1058G	uc010lfj.1	P59
PPT2	9374	32230457	Nonsense	c.229C>A	p.Y42*	uc003nzw.1	P59
PSD	5662	104162257	Missense	c.2146C>G	p.A540G	uc001kvg.1	P59
PTCH2	8643	45065524	Missense	c.2428A>C	p.T806P	uc001cms.1	P59
PTPRB	5787	69220938	Missense	c.5605T>G	p.V1854G	uc001swc.2	P59
RALGPS2	55103	177120882	Missense	c.1294C>G	p.A318G	uc001glz.1	P59
RBM4B	83759	66193265	Missense	c.1155C>G	p.C162W	uc001oja.1	P59
RELT	84957	72783321	Missense	c.1105G>C	p.A314P	uc001otv.1	P59
RFX2	5990	5967270	Missense	c.769T>G	p.L204V	uc002meb.1	P59
RNF152	220441	57634248	Missense	c.841A>T	p.Q143H	uc002lih.1	P59
SCML4	256380	108174684	Missense	c.640T>G	p.V130G	uc010kdf.1	P59
SERINC2	347735	31678427	Missense	c.1190T>G	p.V347G	uc001bst.1	P59
SETD5	55209	9445684	Missense	c.497C>G	p.A21G	uc003brt.1	P59
SETD8	387893	122458184	Missense	c.1082A>C	p.H347P	uc001uew.1	P59
SF3B1	23451	197975079	Missense	c.2146A>G	p.K700E	uc002uu.1	P59

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
SLC35B1	10237	45140157	Missense	c.125G>C	p.R13P	uc002iph.1	P59
SPATS2	65244	48204929	Missense	c.2298T>C	p.Y437H	uc001rud.2	P59
SSPO	23145	149146122	Splice_Site_Del	c.e81_splice_site		uc010lpk.1	P59
SYNE2	23224	63520215	Missense	c.2239A>C	p.N670T	uc001xgl.1	P59
TAF6L	10629	62306382	Missense	c.929G>T	p.W276C	uc009yof.1	P59
THSD7B	80731	137879814	Missense	c.2569G>A	p.E857K	uc002tva.1	P59
TIMD4	91937	156279126	Missense	c.1114G>A	p.D353N	uc003lwh.1	P59
TM4SF19	116211	197538250	Missense	c.377C>G	p.C84W	uc10iad.1	P59
TMPRSS12	283471	49523108	Missense	c.141G>C	p.G32R	uc001rwx.2	P59
UCN3	114131	5406125	Missense	c.666G>C	p.A148P	uc001ihx.1	P59
USP39	10713	85699890	Missense	c.341G>C	p.S102T	uc002sqe.2	P59
WNT10A	80326	219455261	Missense	c.711C>G	p.A83G	uc002vjd.1	P59
WWC2	80014	184419542	Missense	c.1954G>T	p.S59I	uc010irx.1	P59
ZC3H18	124245	87171086	Missense	c.201G>C	p.E18D	uc002fky.1	P59
ZNF264	9422	62408622	Missense	c.619G>A	p.G69E	uc002qob.1	P59
CAPRIN1	4076	34030556	Missense	c.202A>C	p.T5P	uc001mvh.1	P60
CHST11	50515	103675235	Missense	c.878G>C	p.A195P	uc001tkx.1	P60
CLCN3	1182	170793687	Nonsense	c.542C>A	p.Y11*	uc003ish.1	P60
CNN1	1264	11521223	Missense	c.751A>C	p.D196A	uc002mnc.1	P60
COL5A3	50509	9938022	Missense	c.4836A>C	p.T1584P	uc002mmq.1	P60
CUL1	8454	148094596	Missense	c.1326G>C	p.R267P	uc010lpg.1	P60
DGKH	160851	41632171	Nonsense	c.775G>T	p.E252*	uc001uyl.1	P60
FLI1	2313	128133281	Missense	c.252C>G	p.A27G	uc001gem.1	P60
KDM5D	8284	20360855	Missense	c.891C>A	p.Q202K	uc004fug.1	P60
KIF2C	11004	45005103	Nonsense	c.2105G>T	p.E664*	uc001cmg.2	P60
KRTAP19-5	337972	30796183	Missense	c.97C>T	p.R33C	uc002yoi.1	P60
LANC1	10314	211028170	Missense	c.417A>G	p.T105A	uc002ved.1	P60
LGALS8	3964	234768842	Missense	c.375C>G	p.R59G	uc001hxw.1	P60
LOXL2	4017	23273567	Missense	c.851C>T	p.S171L	uc003xdh.1	P60
MAPK14	1432	36103947	Missense	c.397A>G	p.E12G	uc003olp.1	P60
MPDZ	8777	13098980	Missense	c.5985C>G	p.S1978R	uc010rhy.1	P60
MUC2	4583	1083069	Missense	c.12001A>G	p.T3992A	uc001lsx.1	P60
NLGN2	57555	7260977	Missense	c.1716A>C	p.N548T	uc002ggt.1	P60
NUP98	4928	3722354	Missense	c.1660A>C	p.T457P	uc001lyh.1	P60
ODZ2	57451	167554855	Nonsense	c.2850C>A	p.Y950*	uc010jjd.1	P60
PIGT	51604	43487690	Missense	c.1620A>C	p.N516T	uc002xoh.1	P60
PPP2R2C	5522	6431145	Missense	c.248G>C	p.S75T	uc003gia.1	P60
ROR2	4920	93526125	Nonsense	c.2671C>A	p.Y824*	uc004arj.1	P60
SCYL2	55681	99209422	Missense	c.238T>G	p.V63G	uc001ithn.1	P60
SF3B1	23451	197975728	Missense	c.1922G>T	p.R625L	uc002uuue.1	P60
TBC1D25	4943	48288244	Missense	c.388G>A	p.G93R	uc004dk.1	P60
VWC2	375567	49812926	Missense	c.1326C>T	p.T257M	uc003tot.1	P60
ZNF330	27309	142373133	Missense	c.795G>T	p.C192F	uc003iiq.2	P60
10-Sep	151011	109659184	Missense	c.1818T>C	p.I480T	uc002tey.1	P61
ATM	472	107626804	Frame_Shift_Del	c.1787_1788delAA	p.K468fs	uc001pkb.1	P61
BPIL1	80341	31069818	Splice_Site_Del	c.e8_splice_site		uc002wyj.1	P61
C18orf8	29919	19364517	Missense	c.1958T>C	p.F613L	uc010dlt.1	P61
CDK5R2	8941	219533742	Missense	c.1101C>A	p.T319K	uc002vif.1	P61
CES1	1066	54410992	Nonsense	c.970C>T	p.R288*	uc002eil.1	P61
GPR162	27239	6803460	Missense	c.670C>A	p.H45Q	uc001qqw.1	P61
HAPLN4	404037	19229935	Frame_Shift_Del	c.918_919delTG	p.V300fs	uc002nmb.1	P61
IMP3	55272	73719132	Missense	c.1377G>C	p.D145H	uc002bat.2	P61
MICALCL	84953	12328035	Nonsense	c.2095C>T	p.R602*	uc001mk.1	P61
MKRN3	7681	21362042	Missense	c.496C>T	p.P7L	uc001ywh.2	P61
SF3B1	23451	197975079	Missense	c.2146A>G	p.K700E	uc002uuue.1	P61
SLC6A5	9152	20632917	Missense	c.2594T>A	p.I774N	uc001mqd.1	P61
SPOCK1	6695	136342286	Missense	c.1467G>C	p.D426H	uc003lbo.1	P61
SPP2	6694	234632271	Missense	c.348G>A	p.R88Q	uc002vvk.1	P61
ZNF527	84503	42571176	Missense	c.496G>A	p.A129T	uc010efk.1	P61
ALMS1	7840	73466540	In_frame_Del	c.147_152delGGA	p.EE27del	uc002sje.1	P62
DUOX2	50506	43190176	Missense	c.1110C>G	p.P303A	uc010bea.1	P62
RFT1	91869	53113134	Missense	c.1024C>G	p.A326G	uc003dgj.1	P62
TP53	7157	7517846	Missense	c.1011C>T	p.R273C	uc002gim.2	P62
ABRA	137735	107851012	Missense	c.637G>A	p.G195S	uc003ym.2	P63
APAF1	317	97595380	Missense	c.2417C>G	p.H614D	uc001tfz.1	P63
C9orf86	55684	138853337	Missense	c.1896C>G	p.A480G	uc004cj.1	P63
COL4A2	1284	109956830	Nonsense	c.4759C>A	p.Y1490*	uc001vqx.1	P63
CSMD3	114788	113632184	Missense	c.4615A>T	p.S1486C	uc003ynu.1	P63
DSG4	147409	27226246	Missense	c.1085G>T	p.W317L	uc002kwr.1	P63
GAS2L1	10634	28034328	Missense	c.432C>G	p.A78G	uc003afa.1	P63
GPR113	165082	26390905	Missense	c.1015G>C	p.R338P	uc002rhe.2	P63
GPR135	64582	59001142	Missense	c.671G>C	p.A186P	uc010apj.1	P63
GPR172A	79581	145554721	Missense	c.918C>T	p.P254L	uc003zcc.1	P63
GRM3	2913	86253850	Missense	c.1905C>T	p.A269V	uc003uid.1	P63

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
KRT26	353288	36181001	Missense	c.501C>T	p.T152I	uc002hv.f.1	P63
LRP1	4035	55876248	Missense	c.9279G>C	p.G2938A	uc001snd.1	P63
MRM1	79922	32032797	Missense	c.660G>C	p.V149L	uc002hne.1	P63
PRPF8	10594	1524616	Missense	c.3283C>T	p.R1057W	uc002fte.1	P63
RBBP6	5930	24480693	Frame_Shift_Del	c.2039_2039delA	p.R333fs	uc002dmh.1	P63
RLTPR	146206	66238135	Frame_Shift_Del	c.604_604delT	p.Y162fs	uc002etn.1	P63
SEMA6D	80031	45848127	Missense	c.2183C>A	p.P608H	uc010bek.1	P63
THBD	7056	22977192	Missense	c.1110C>G	p.A317G	uc002wss.1	P63
ZNF449	203523	134308854	Frame_Shift_Del	c.285_285delA	p.N49fs	uc004eys.1	P63
ANKRD13B	124930	24959220	Missense	c.454C>G	p.A114G	uc002hei.1	P64
ANP32D	23519	47152794	Missense	c.80G>A	p.S27N	uc001rrq.1	P64
DLAT	1737	111419435	Missense	c.1824A>G	p.I389V	uc001pmo.2	P64
DUSP27	92235	165353302	Missense	c.319A>C	p.T107P	uc001geb.1	P64
EIF5	1983	102871991	Missense	c.563A>T	p.Y14F	uc001ymq.1	P64
ELOVL6	79071	111190493	Splice_Site_Del	c.e4_splice_site		uc003iaa.1	P64
ERBB2IP	55914	65386515	Missense	c.3658G>A	p.E1201K	uc010iwx.1	P64
FSCB	84075	44044152	Missense	c.2057G>A	p.A597T	uc001wvn.1	P64
GRM7	2917	7595194	Missense	c.1750A>C	p.K534T	uc003bql.1	P64
HIPK3	10114	33317497	Missense	c.1724G>C	p.G485A	uc001mul.1	P64
KCNJ16	3773	65616093	De_novo_Start_OutOfFrame	c.272_273insT		uc002jin.1	P64
MDF1	4188	41721911		c.475C>A	p.P49Q	uc003oqp.2	P64
NRP2	8828	206300937	Nonsense	c.1859C>A	p.Y356*	uc002vaw.1	P64
PER2	8864	238834304	Nonsense	c.1683C>A	p.Y482*	uc002vyc.1	P64
POP7	10248	100142684	Missense	c.557G>C	p.A99P	uc003uwh.2	P64
SETDB1	9869	149190120	Missense	c.2260A>G	p.K715E	uc001evu.1	P64
SLC7A4	6545	19715788	Missense	c.382T>A	p.F105Y	uc002zud.1	P64
SPTBN2	6712	66232330	Missense	c.1280A>G	p.E403G	uc001ojd.1	P64
SRGAP2	23380	204633613	Missense	c.863T>C	p.V177A	uc001hd.y.1	P64
TM7SF2	7108	64638857	Missense	c.1360G>A	p.V255M	uc001ocv.1	P64
TNK2	10188	197093554	Missense	c.986C>A	p.R281S	uc003fvt.1	P64
USP34	9736	61369506	Missense	c.4581A>T	p.D1520V	uc002sbe.1	P64
VIPR2	7434	158595268	Missense	c.441A>C	p.K85N	uc003woh.1	P64
ACAN	176	87196231	Missense	c.2603A>C	p.E743D	uc002bmy.1	P65
BID	637	16602132	Missense	c.808A>C	p.T162P	uc002znc.1	P65
C9orf93	203238	15961794	Missense	c.4256T>C	p.M1314T	uc003zmd.1	P65
FLG2	388698	150594244	Missense	c.2715A>C	p.Y881S	uc001ezw.2	P65
GAN	8139	79953652	Missense	c.1165C>G	p.L341V	uc002fgo.1	P65
GRK7	131890	143009376	Missense	c.1334A>G	p.D417G	uc003euf.1	P65
HCN1	348980	45432433	Missense	c.1173C>T	p.A383V	uc003jok.1	P65
MGA	23269	39815981	Frame_Shift_Del	c.4408_4408delT	p.A1409fs	uc001zoh.1	P65
NOTCH1	4851	138510470	Frame_Shift_Del	c.7541_7542delCT	p.P2514fs	uc004chz.1	P65
PLXNA2	5362	206282243	Missense	c.4867G>A	p.R1370H	uc001hg.z.1	P65
PTPRH	5794	60400300	Missense	c.2028A>G	p.T663A	uc002ajq.1	P65
RBM6	10180	50070866	Missense	c.2130C>T	p.S666F	uc003cyc.1	P65
RIMKLB	57494	8817563	Frame_Shift_Ins	c.1328_1329insC	p.E359fs	uc001quu.2	P65
RPLP0	6175	119121066	Missense	c.676A>T	p.I147F	uc001txp.1	P65
SLITRK3	22865	166388975	Missense	c.2782C>A	p.P780T	uc003fej.2	P65
SPEN	23013	16128464	Nonsense	c.3346G>T	p.E1048*	uc001axk.1	P65
SPERT	220082	45185415	Missense	c.334C>T	p.A85V	uc001van.1	P65
TP53	7157	7517845	Missense	c.1012G>A	p.R273H	uc002gim.2	P65
ZC3H12B	340554	64639529	Nonsense	c.2202C>A	p.Y731*	uc010nko.1	P65
ZFHX3	463	71403304	Splice_Site_SNP	c.e6_splice_site		uc002fck.1	P65
ARID1B	57492	157264338	Missense	c.1852A>C	p.Y567S	uc003qqn.1	P66
ASTE1	28990	132215847	Missense	c.2066G>T	p.S620I	uc010htm.1	P66
C14orf43	91748	73263919	De_novo_Start_OutOfFrame	c.1714C>G		uc001xos.1	P66
CD2BP2	10421	30272477	Missense	c.774C>G	p.A174G	uc002dxr.1	P66
CHRNB4	1143	76714907	Missense	c.245C>T	p.R45C	uc002bed.1	P66
CNOT3	4849	59339217	Missense	c.489G>A	p.D60N	uc002qdi.1	P66
COL4A3	1285	227867981	Missense	c.3638G>A	p.R1159H	uc002vom.1	P66
CPS1	1373	211175209	Nonsense	c.1843C>A	p.Y588*	uc010fur.1	P66
DST	667	56579872	Missense	c.6010T>A	p.Y1968N	uc003pdः.2	P66
FLNC	2318	128257948	Nonsense	c.230C>A	p.Y7*	uc003vnz.2	P66
FTH1	2495	61489465	Missense	c.448G>A	p.M71I	uc001nsu.1	P66
GFI1B	8328	134853570	Missense	c.555T>C	p.V135A	uc004ccg.1	P66
GJC2	57165	226413328	Missense	c.1421G>A	p.G416R	uc001hsk.1	P66
KLF9	687	72218065	Missense	c.1329C>G	p.A12G	uc004aht.1	P66
MAEL	84944	165225305	Missense	c.163G>C	p.R31P	uc001gdy.1	P66
MANBA	4126	103811592	Missense	c.1224T>C	p.L375P	uc003hwg.1	P66
MYD88	4615	38157645	Missense	c.794T>C	p.L265P	NM_002468	P66
PHLDB1	23187	118020040	Missense	c.3412G>T	p.R1020L	uc001ptr.1	P66
PLEKHH1	57475	67118588	Missense	c.3476C>G	p.L1112V	uc001xjl.1	P66
PLEKHN1	84069	898186	In_frame_Del	c.1276_1278delGC	p.R414_415RT>P	uc001ace.1	P66
SCN8A	6334	50401812	Nonsense	c.2029C>A	p.Y617*	uc001ryw.1	P66
SF3A2	8175	2199165	In_frame_Del	c.1137_1157delCC	p.PAPGVHP360del	uc002lvg.1	P66

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SIRPA	140885	1851282	Missense	c.1087A>C	p.T360P	uc002wft.1	P66
SMC3	9126	112351888	Missense	c.3193C>T	p.L1023F	uc001kze.1	P66
SMYD1	150572	88168501	Missense	c.322C>G	p.A107G	uc002ssr.1	P66
TNRC6A	27327	24649089	Missense	c.134A>G	p.K7R	uc002dmm.1	P66
UPK1A	11045	40856258	Missense	c.439A>C	p.T147P	uc010eeh.1	P66
ZNF711	7552	84409954	Splice_Site_SNP	c.e8_splice_site		uc004eeq.1	P66
AATK	9625	76708382	Frame_Shift_Ins	c.3911_3912insC	p.P1277fs	uc010dia.1	P67
ACTL8	81569	18022392	Missense	c.482C>T	p.T101M	uc001bat.1	P67
AHDC1	27245	27746594	Missense	c.5589C>G	p.C1540W	uc009vsy.1	P67
CD22	933	40518809	Missense	c.520C>T	p.P148L	uc010edt.1	P67
CDH15	1013	87786248	Missense	c.1827A>C	p.K584Q	uc002fmr.1	P67
CDH9	1007	26926414	Missense	c.1439C>T	p.H424Y	uc003jgs.1	P67
CNBD1	168975	88318317	Missense	c.680C>A	p.T211K	uc003ydy.2	P67
CREBBP	1387	3718097	Missense	c.7156C>G	p.Q2318E	uc002cvv.1	P67
CSMD3	114788	113416867	Missense	c.7191C>A	p.D2344E	uc003ynu.1	P67
DUSP2	1844	96173628	Missense	c.808G>A	p.G241D	uc002svk.2	P67
ERAL1	26284	24206186	Missense	c.18C>G	p.A3G	uc002hcy.1	P67
JAG2	3714	104685720	Missense	c.2526A>C	p.T708P	uc001ygg.1	P67
MUT	4594	49516005	Missense	c.2084G>A	p.R610H	uc003ozg.2	P67
MYBL2	4605	41743895	Missense	c.387G>C	p.A58P	uc002xlb.1	P67
MYD88	4615	38157263	Missense	c.695T>C	p.M232T	NM_002468	P67
NBEA	26960	34415190	Missense	c.439T>C	p.I78T	uc001uvb.1	P67
PBX2	5089	32265621	Frame_Shift_Del	c.321_321delG	p.G17fs	uc003oav.1	P67
PVRL2	5819	50073438	In_frame_Del	c.1551_1553delGA	p.R391del	uc002ozv.1	P67
SI	6476	166265856	Missense	c.756C>T	p.R232C	uc003fei.1	P67
SLC44A3	126969	95129325	Missense	c.1641A>G	p.K512E	uc001dgv.2	P67
SMCHD1	23347	2695692	Missense	c.2032G>A	p.V615I	uc002klm.2	P67
SYT7	9066	61047911	Missense	c.1102C>T	p.R366W	uc009yrn.1	P67
TRIM11	81559	226649486	Missense	c.1205C>G	p.A317G	uc001hss.1	P67
ZNF697	90874	119966957	Missense	c.1646A>C	p.H511P	uc001ehy.1	P67
ABI3BP	25890	101954474	Missense	c.2901G>A	p.D946N	uc003dun.1	P68
C11orf41	25758	33587964	Missense	c.4406G>A	p.A1428T	uc001mup.2	P68
CLASP1	23332	121861233	Missense	c.3699C>A	p.H1103Q	uc002tnc.1	P68
CTTNBP2NL	55917	112800515	Missense	c.1046C>T	p.P293L	uc001ebx.1	P68
DMXL1	1657	118512389	Missense	c.3149A>G	p.T990A	uc010jcl.1	P68
DOCK8	81704	410429	Missense	c.3981C>T	p.A1290V	uc003zgf.1	P68
GOLGA3	2802	131900009	Missense	c.1035A>G	p.E159G	uc001ukz.1	P68
KRT83	3889	51001206	Missense	c.244G>A	p.A61T	uc001saf.2	P68
LRRC4C	57689	40093863	Missense	c.2520T>C	p.S186P	uc001mxa.1	P68
MUC2	4583	1083430	Missense	c.12362C>A	p.T4112N	uc001lsx.1	P68
OR13C8	138802	106371360	Missense	c.91A>G	p.I31V	uc004bcc.1	P68
RIMS4	140730	42818349	Missense	c.653G>C	p.R218P	uc010ggu.1	P68
RPUSD2	27079	38651347	Missense	c.859G>A	p.A287T	uc001zmd.1	P68
RXFP1	59350	159774068	Missense	c.1043C>A	p.L321M	uc003ipz.1	P68
SDC1	6382	20267419	Missense	c.562C>A	p.A88D	uc002rdo.1	P68
SKA3	221150	20633928	Splice_Site_SNP	c.e5_splice_site		uc001unt.1	P68
TAS2R41	259287	142885224	Missense	c.137T>C	p.M46T	uc003wdc.1	P68
TERF2IP	54386	74239345	Missense	c.161G>T	p.V22L	uc002fet.1	P68
TRYX3	136541	141601831	Splice_Site_SNP	c.e2_splice_site		uc003vbx.1	P68
ALS2CR8	79800	203527027	Missense	c.762C>A	p.T161N	uc002uzo.2	P69
ARRDC1	92714	139628914	Missense	c.952C>T	p.P293L	uc004cnp.1	P69
CALHM1	255022	105208073	Missense	c.563C>G	p.S142R	uc001kxe.1	P69
CCNB3	85417	50107426	Missense	c.4170A>C	p.Q1291P	uc004dox.2	P69
CPXM1	56265	2726933	Missense	c.519G>A	p.G152D	uc002wgu.1	P69
DICER1	23405	94630229	Missense	c.5295G>A	p.E1705K	uc001ydw.2	P69
DLGAP5	9787	54695137	Missense	c.1946G>C	p.A577P	uc001xbx.1	P69
DOCK7	85440	62892051	Missense	c.356G>A	p.E108K	uc001daq.1	P69
FAM135B	51059	139224514	Missense	c.373T>A	p.F1187L	uc003yuy.1	P69
GRB14	2888	165112505	Missense	c.933G>C	p.R131P	uc002ucl.1	P69
ITGA9	3680	37801572	Missense	c.2941C>T	p.T963M	uc003chd.1	P69
MED1	5469	34817880	Missense	c.4332T>C	p.S1374P	uc002hrv.2	P69
MIIP	60672	12011694	Missense	c.745T>C	p.S189P	uc001ato.1	P69
NHEDC1	150159	104047234	Missense	c.1225T>G	p.I368S	uc003hww.1	P69
PAK7	57144	9572897	Missense	c.625C>T	p.P27L	uc002wml.2	P69
PXN	5829	119138181	Missense	c.940G>A	p.E20K	uc001txu.2	P69
ABCC3	8714	46088335	Nonsense	c.259C>A	p.Y63*	uc002isl.1	P70
ACLY	47	37297388	Missense	c.2032G>C	p.G676A	uc002hyi.1	P70
AGTR1	185	149942251	Missense	c.1185C>A	p.L247I	uc003ewg.1	P70
ALMS1	7840	73532015	Missense	c.4967G>T	p.S1619I	uc002sje.1	P70
APOB	338	21083778	Frame_Shift_Ins	c.9594_9595insA	p.T3156fs	uc002red.1	P70
ATP2B2	491	10362785	Missense	c.2760T>G	p.V814G	uc003bvt.1	P70
CACNA1G	8913	46031978	Missense	c.3821G>A	p.R1150Q	uc002irk.1	P70
CERCAM	51148	130236577	Missense	c.1797G>A	p.V467M	uc004buz.2	P70
DOK3	79930	176862778	In_frame_Del	c.1026_1028delCT	p.L289del	uc003mhi.2	P70

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
FBXL21	26223	135304105	Missense	c.539T>C	p.V173A	uc010jec.1	P70
HIST1H4F	8361	26348931	Missense	c.299G>A	p.G100D	uc003nhe.1	P70
KIAA1244	57221	138697762	Missense	c.6086A>G	p.Y2029C	uc003qhu.2	P70
KIF26A	26153	103688444	Missense	c.628G>A	p.A210T	uc001yos.2	P70
KIF26B	55083	243916439	Missense	c.3971G>C	p.Q1177H	uc001ibf.1	P70
NR2F2	7026	94678458	Missense	c.1173A>G	p.S198G	uc002btq.1	P70
RAG2	5897	36572292	Missense	c.191G>A	p.M11	uc001mwv.2	P70
RIF1	55183	151981368	Missense	c.458A>G	p.N110D	uc002txm.1	P70
ROBO2	6092	77696868	Missense	c.2399C>T	p.R586W	uc003dpv.2	P70
SELO	83642	48991188	Missense	c.1130T>G	p.Y358D	uc003bjx.1	P70
TAF4B	6875	22149232	Missense	c.2378T>G	p.V630G	uc002kv.2	P70
TAF7L	54457	100434548	Missense	c.154G>A	p.D48N	uc004ehb.1	P70
TMEM79	84283	154528792	Splice_Site_Del	c.e4_splice_site		uc001foe.1	P70
ZBTB10	65986	81562423	Missense	c.1421T>G	p.C275G	uc003ybx.2	P70
ABI3BP	25890	102066342	Frame_Shift_Del	c.1174_1174delT	p.F363fs	uc003dup.2	P71
FASN	2194	77639393	Nonsense	c.2790C>A	p.Y891*	uc002kdu.1	P71
FOXJ3	22887	42549329	Missense	c.335G>T	p.C8F	uc001che.1	P71
SUSD3	203328	94877910	Missense	c.148C>A	p.P38T	uc004atb.1	P71
BPII3	128859	31093481	Missense	c.1187A>G	p.N396S	uc002wyk.1	P72
C12orf5	57103	4331955	Missense	c.79T>C	p.L217S	uc001qmp.1	P72
CELSR1	9620	45308699	Missense	c.3033C>G	p.N1011K	uc003bhw.1	P72
CFC1B	653275	131072730	Missense	c.593T>G	p.W68G	uc002tro.1	P72
CSMD1	64478	2953627	Missense	c.7052C>A	p.T2221K	uc010lrh.1	P72
DTNA	1837	30711720	Missense	c.1863C>G	p.A621G	uc010dmn.1	P72
DYNC1L12	1783	65319629	Missense	c.1150A>C	p.Q373H	uc002eqb.1	P72
DYRK1B	9149	45008557	Missense	c.1808T>C	p.S510P	uc002omj.1	P72
ELF1	1997	40416032	Missense	c.787T>C	p.S187P	uc001uxr.1	P72
FAM179A	165186	29103252	Missense	c.2234C>T	p.A628V	uc010ezl.1	P72
FOXJ2	55810	8091870	Missense	c.2028T>C	p.S315P	uc001qtu.1	P72
GFM1	85476	159866794	Missense	c.1633A>G	p.E509G	uc003fce.1	P72
IFT122	55764	130715978	Missense	c.3403C>G	p.A1066G	uc003eml.1	P72
IGFN1	91156	199452330	Missense	c.1673C>T	p.R301W	uc001gwc.1	P72
KCNS2	3788	99510478	Missense	c.1445G>C	p.W365C	uc003yin.1	P72
LYPD5	284348	48994512	Missense	c.533C>G	p.S151C	uc002oxm.2	P72
MAGEA8	4107	148774495	Missense	c.1006C>T	p.A264V	uc004fdw.1	P72
MESP2	145873	88121151	In_frame_Del	c.559_570delGGG	p.GQGQ199del	uc002bon.1	P72
METTL13	51603	170019650	Missense	c.648T>G	p.L101V	uc001ghz.1	P72
PLCD3	113026	40550913	Nonsense	c.1348C>T	p.Q412*	uc002iib.1	P72
PRKCI	5584	171463881	Missense	c.572C>T	p.R112C	uc003fgs.2	P72
PTTG1	9232	159781905	Missense	c.53C>A	p.T3N	uc003lyj.1	P72
RAB21	23011	70450651	Missense	c.484C>G	p.Q78E	uc001swt.1	P72
RPS15	6209	1391458	Missense	c.576G>C	p.K152N	uc002lsq.1	P72
TBC1D25	4943	48304293	Missense	c.2164A>C	p.T685P	uc004dka.1	P72
TNK2	10188	197079875	Missense	c.2025C>G	p.A627G	uc003fv.1	P72
TOPBP1	11073	134821752	Missense	c.3393C>T	p.S1016F	uc003eps.1	P72
TP53	7157	7519095	Splice_Site_SNP	c.e5_splice_site		uc002gim.2	P72
12-Sep	124404	4767885	Missense	c.1080G>A	p.A331T	uc002cxq.1	P73
ADAMTS7	11173	76838856	Missense	c.5234G>T	p.A1675S	uc002bej.2	P73
ASB12	142689	63361600	Missense	c.690G>C	p.R219P	uc004dvq.1	P73
ATM	472	107707431	Missense	c.7951A>T	p.Q2522H	uc001pkb.1	P73
ATM	472	107721712	Nonsense	c.8836T>G	p.Y2817*	uc001pkb.1	P73
ATP1A1	476	116742907	Missense	c.2564G>A	p.A756T	uc001ege.1	P73
ATP8B3	148229	1747166	Missense	c.2086G>C	p.V618L	uc002ltw.1	P73
BRD8	10902	137504292	Splice_Site_SNP	c.e26_splice_site		uc003lcf.1	P73
C14orf43	91748	73263933	Missense	c.2926A>T	p.T715S	uc001xot.1	P73
CHD5	26038	6129398	Missense	c.1604C>T	p.P502S	uc001amb.1	P73
CNTN5	53942	99675173	Missense	c.2794C>T	p.R819C	uc001pgg.1	P73
DAPK1	1612	89501868	Missense	c.2678A>T	p.E847V	uc004apc.1	P73
DOK6	220164	65659552	Missense	c.1139C>T	p.R317W	uc002lkl.1	P73
ERBB2IP	55914	65385399	Missense	c.2542C>T	p.P829S	uc010iwx.1	P73
ESPL1	9700	51949811	Missense	c.909G>A	p.S273N	uc001scck.2	P73
FAM92A1	137392	94809636	Missense	c.908C>G	p.Q269E	uc010maq.1	P73
FAT4	79633	126462093	Missense	c.5077G>A	p.A1693T	uc003if.2	P73
FCER1A	2205	157542410	Missense	c.439C>G	p.L114V	uc001ftq.1	P73
GABRA5	2558	24765119	Missense	c.961G>A	p.G208S	uc001zbd.1	P73
GJC3	349149	99364644	Missense	c.536C>T	p.T179I	uc003usg.1	P73
IGSF11	152404	120127650	Missense	c.815A>G	p.T190A	uc003ebw.1	P73
ITK	3702	156570945	Missense	c.395G>T	p.A105S	uc003lwo.1	P73
KCNK18	338567	118959115	Missense	c.470C>T	p.T157I	uc001ldc.1	P73
LMLN	89782	199171558	Missense	c.91C>T	p.P12S	uc003fy.1	P73
LRIT2	340745	85975249	Missense	c.16C>T	p.S3L	uc001kcy.1	P73
NVL	4931	222554957	Missense	c.1035C>G	p.A331G	uc001hok.1	P73
OBSCN	84033	226573385	Missense	c.1435G>C	p.R4770P	uc009xez.1	P73
PHF3	23469	64471502	Frame_Shift_Del	c.3375_3381delAA	p.N1117fs	uc003pep.1	P73

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
RHBDD3	25807	27991524	Missense	c.464T>G	p.V31G	uc003aeq.1	P73
RSAD2	91543	6944657	Missense	c.785G>A	p.A217T	uc002qyp.1	P73
SLC4A11	83959	3157652	Missense	c.2120T>G	p.V691G	uc002wig.1	P73
TNFAIP2	7127	102662697	In_frame_Del	c.281_283delGAA	p.K54del	uc001ymm.1	P73
TSHZ2	128553	51303553	Missense	c.1105C>T	p.T50M	uc002xwo.2	P73
TSPAN33	340348	128588805	Missense	c.381A>T	p.K51M	uc003vop.1	P73
UGT1A4	54657	234293054	Missense	c.878C>G	p.N283K	uc002vux.1	P73
USH2A	7399	214078036	Missense	c.9678A>T	p.K3097N	uc001hku.1	P73
USP19	10869	49124422	Nonsense	c.2990C>A	p.Y943*	uc003cvz.2	P73
A2M	2	9145502	Nonsense	c.1415C>A	p.Y434*	uc001qvk.1	P74
ABCC6	368	16167016	Missense	c.3308A>C	p.I1091L	uc002den.2	P74
ADAM15	8751	153297363	Missense	c.1840A>C	p.Q580P	uc001fgr.1	P74
C11orf88	399949	110892001	Missense	c.295C>A	p.L99I	uc009yyd.1	P74
C15orf2	23742	22472528	Missense	c.895A>C	p.I141L	uc001ywo.1	P74
COL11A2	1302	33261505	Missense	c.1055A>T	p.E276V	uc003ocx.1	P74
CYP27C1	339761	127669546	Missense	c.685C>A	p.T185K	uc002itod.2	P74
DERL2	51009	5330180	Missense	c.42A>G	p.E9G	uc002gcc.1	P74
FAM103A1	83640	81449669	Missense	c.388A>G	p.D68G	uc002bjl.1	P74
FAM151B	167555	79873378	Missense	c.945C>A	p.Q268K	uc003kgv.1	P74
FUT7	2529	139045459	Missense	c.1402C>G	p.L185V	uc004ckq.2	P74
HECTD1	25831	30683882	Missense	c.3002G>C	p.R838P	uc001wrc.1	P74
KLHL31	401265	53624918	Missense	c.1483C>G	p.P448A	uc003pcb.2	P74
MLL3	58508	151495360	Missense	c.9773A>C	p.Q3185P	uc003wla.1	P74
OLFML3	56944	114325104	Nonsense	c.520C>A	p.Y137*	uc001eer.1	P74
PTCH1	5727	97260245	Nonsense	c.3227C>A	p.Y1013*	uc004avk.2	P74
RBKS	64080	27919537	Missense	c.426G>C	p.A139P	uc002rl.1	P74
RELT	84957	72783932	Missense	c.1364G>A	p.G400E	uc001otv.1	P74
RNF10	9921	119457061	Missense	c.547A>C	p.N22H	uc001typ.2	P74
SEMA3A	10371	83448699	Missense	c.1841T>G	p.V509G	uc003uhz.1	P74
SLC25A33	84275	9562832	Missense	c.939C>G	p.A239G	uc001apw.1	P74
TP53	7157	7520080	Missense	c.526T>G	p.L111R	uc002gim.2	P74
CA10	56934	47065998	Missense	c.1556C>T	p.R274C	uc002itv.2	P75
CRAMP1L	57585	1643068	Missense	c.687G>C	p.R112P	uc002cme.1	P75
DUS3L	56931	5740592	Missense	c.574A>C	p.T176P	uc002mdc.1	P75
DYNC2H1	79659	102844538	Missense	c.12825G>T	p.L4227F	uc001phn.1	P75
ELN	2006	73104225	Missense	c.1016G>C	p.A309P	uc003tzw.1	P75
EPM2A	7957	145990417	Missense	c.1181C>T	p.A275V	uc003qkw.1	P75
GAP43	2596	116878018	Missense	c.980C>A	p.Q203K	uc003ebr.1	P75
ITPKB	3707	224990032	Frame_Shift_Ins	c.1786_1787insG	p.E584fs	uc001hqg.1	P75
KIAA0182	23199	84248567	Missense	c.1570C>G	p.A499G	uc002fix.1	P75
NFASC	23114	203214793	Frame_Shift_Del	c.2150_2150delG	p.G651fs	uc001hbj.1	P75
PRR21	643905	240630159	Frame_Shift_Del	c.901_914delGCC	p.A301fs	uc002vys.1	P75
SLAMF1	6504	158873631	Missense	c.735G>A	p.R130H	uc001fvl.2	P75
TFEB	7942	41761846	Missense	c.1047G>A	p.R318H	uc003oqu.1	P75
ADAMTS19	171019	129047739	Missense	c.2674G>A	p.G892S	uc003kvb.1	P76
BID	637	16602132	Missense	c.808A>C	p.T162P	uc002znc.1	P76
C17orf71	55181	54642204	Missense	c.52C>G	p.P4A	uc002ixi.1	P76
CASKIN1	57524	2170623	Missense	c.2779G>C	p.R916P	uc010bsg.1	P76
CHMP7	91782	23169973	Missense	c.1361A>G	p.D238G	uc003xdc.2	P76
COPG	22820	130478921	Missense	c.2689G>C	p.E863D	uc003els.1	P76
DLG5	9231	79265503	Missense	c.1691C>T	p.R541W	uc001jzk.1	P76
GALNT3	2591	166319480	Missense	c.1916A>G	p.K510R	uc010fph.1	P76
KLHL11	55175	37274800	Missense	c.356C>G	p.A117G	uc002hyf.1	P76
LRRIQ1	84125	84024438	Missense	c.3402A>T	p.K1097N	uc001tac.1	P76
MRC2	9902	58097890	Missense	c.1302G>T	p.L300F	uc002jad.1	P76
NEU4	129807	242406849	Nonsense	c.1744C>A	p.Y431*	uc002wcn.1	P76
NINJ2	4815	544793	Nonsense	c.527C>T	p.R146*	uc001qil.1	P76
PCDHA8	56140	140202654	Missense	c.1564C>T	p.P522S	uc003lhs.1	P76
RGS9	8787	60594848	Missense	c.679T>G	p.V190G	uc002jfe.1	P76
SSPO	23145	149124440	Missense	c.6583G>A	p.G2195S	uc010lpk.1	P76
STAB1	23166	52529348	Splice_Site_SNP	c.e52_splice_site		uc003dej.1	P76
STOX1	219736	70314588	Missense	c.1030G>A	p.V344I	uc001joq.1	P76
TAOK1	57551	24849472	Missense	c.1204A>G	p.H337R	uc002hdz.1	P76
TBC1D23	55773	101517661	Missense	c.1634A>G	p.K543E	uc003dtt.1	P76
TBC1D28	254272	18483233	Missense	c.590G>A	p.V60I	uc002gud.2	P76
TP53	7157	7518996	Missense	c.772A>T	p.H193L	uc002gim.2	P76
TP53AIPI1	63970	128312725	Missense	c.409C>T	p.L67F	uc001qex.1	P76
VPS41	27072	38764580	Missense	c.1475G>T	p.W483C	uc003tgj.1	P76
BMPER	168667	33943462	Missense	c.629G>T	p.V86L	uc003tdw.1	P77
CSMD1	64478	3876895	Missense	c.940A>G	p.T184A	uc010lrh.1	P77
DENND1A	57706	125184134	Missense	c.2661C>T	p.P810S	uc004bnz.1	P77
DHX37	57647	124031223	In_frame_Del	c.601_603delGAG	p.E168del	uc001ugy.1	P77
DOCK6	57572	11222606	Missense	c.705C>G	p.L222V	uc002mqs.2	P77
DSP	1832	7500957	Missense	c.457G>A	p.G60S	uc003mfp.1	P77

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FAT1	2195	187865514	Missense	c.2650A>T	p.E821V	uc003izf.1	P77
IL12RB2	3595	67589273	Missense	c.1811G>A	p.G391R	uc001ddu.1	P77
IRAK4	51135	42466478	Missense	c.1322A>G	p.K400E	uc001rnu.2	P77
MAN1C1	57134	25952568	Nonsense	c.1171C>T	p.R281*	uc001bkm.2	P77
NBPF1	55672	16781708	Frame_Shift_Del	c.2112_2112delC	p.D408fs	uc009vos.1	P77
NPL	80896	181030157	Missense	c.168G>A	p.G10S	uc009wyb.1	P77
PRKAR1B	5575	717535	Missense	c.240G>A	p.R45H	uc003siu.1	P77
PRR21	643905	240630790	Frame_Shift_Del	c.256_283delAGT	p.S86fs	uc002vys.1	P77
PSD3	23362	18774161	Missense	c.596T>C	p.S165P	uc003wza.1	P77
PTK2B	2185	27352517	Missense	c.2504G>A	p.G566R	uc003xfn.1	P77
RAMP3	10268	45164003	Frame_Shift_Del	c.112_112delG	p.L17fs	uc003tnb.1	P77
SCN7A	6332	167037099	Nonsense	c.673G>A	p.W182*	uc002udu.1	P77
TAF6	6878	99543163	Missense	c.1984C>T	p.S616L	uc003uth.1	P77
UCK2	7371	164141791	Missense	c.788T>A	p.Y203N	uc001gdp.1	P77
WARS	7453	99889892	Missense	c.694A>C	p.K204Q	uc001yhf.1	P77
C6orf1	221491	34322597	Missense	c.744C>A	p.T51N	uc003ojf.1	P78
CPNE7	27132	88189378	Missense	c.1760A>G	p.I544V	uc002fnp.1	P78
DAPK1	1612	89511703	Missense	c.4035C>A	p.D1299E	uc004apc.1	P78
DLG5	9231	79271650	Missense	c.1502C>T	p.R478W	uc001jzk.1	P78
FAT3	120114	92173109	Missense	c.7299C>T	p.R2428W	uc001pdj.2	P78
GABRA2	2555	46007001	Missense	c.1178C>T	p.H169Y	uc003gxc.2	P78
GRK4	2900	120338435	Missense	c.2388G>A	p.V701M	uc001pxn.2	P78
HDGFRP2	84717	4448957	Missense	c.1424C>T	p.P444L	uc002mao.1	P78
IL28B	282617	44426941	Missense	c.218C>T	p.R72C	uc002oks.1	P78
MAOB	4129	43587945	Missense	c.232C>G	p.A19G	uc004dfz.2	P78
MED12	9968	70255978	Missense	c.329G>A	p.G44S	uc004dy.1	P78
SYTL2	54843	85096119	Splice_Site_SNP	c.e8_splice_site		uc001ppb.1	P78
WDR7	23335	52597686	Missense	c.3185T>C	p.C992R	uc002ljk.1	P78
WDR72	256764	51812596	Frame_Shift_Del	c.85_85delG	p.A15fs	uc002acj.2	P78
AKAP8L	26993	15390730	Missense	c.104G>A	p.S2N	uc002nav.1	P79
ALDH5A1	7915	24623476	Missense	c.896G>A	p.V290M	uc003nef.1	P79
C1QL1	10882	40400864	Missense	c.307A>C	p.T27P	uc002ihv.1	P79
DOCK5	80005	25205517	Frame_Shift_Ins	c.519_520insGG	p.R128fs	uc003xeg.1	P79
EPPK1	83481	145015572	Missense	c.3851C>G	p.L1255V	uc003zaa.1	P79
FAM120A	23196	95254365	Missense	c.372G>C	p.R116P	uc004atw.1	P79
KCNU1	157855	36761243	Frame_Shift_Del	c.244_244delA	p.K53fs	uc010lvw.1	P79
KIAA1524	57650	109784542	Missense	c.598G>C	p.S110T	uc003dxb.2	P79
MED12L	116931	152391387	Missense	c.1985G>T	p.K649N	uc003eyp.1	P79
PFN1	5216	4790826	Missense	c.303G>A	p.R56Q	uc002gaa.1	P79
PLXNA1	5361	128219828	Missense	c.3597T>G	p.V1198G	uc003ejg.1	P79
PODXL	5420	130891570	In_frame_Del	c.342_347delGTC	p.28_30PSP>P	uc003vqw.2	P79
PPFIA2	8499	80179892	Read-through	c.3935A>C	p.*1258C	uc001szo.1	P79
RFTN2	130132	198206795	Missense	c.1012G>A	p.G204R	uc002uuu.2	P79
SPG20	23111	35807294	Missense	c.768C>A	p.P225Q	uc001uvm.1	P79
STAB2	55576	102624878	Missense	c.4361G>C	p.G1392A	uc001tjw.1	P79
TNS3	64759	47375185	Missense	c.1950A>G	p.N528S	uc003tnv.1	P79
ZMAT5	55954	28464404	Missense	c.549C>A	p.L100I	uc003agm.1	P79
BAI3	577	69405721	Missense	c.881G>A	p.G145R	uc003pev.2	P80
CCDC62	84660	121852039	Missense	c.1538A>T	p.S465C	uc001udc.1	P80
COL5A2	1290	189607103	Missense	c.4713G>A	p.V1480M	uc002uqk.1	P80
DPP9	91039	4653620	Missense	c.1156G>T	p.W293L	uc002mba.1	P80
KAL1	3730	8461076	Missense	c.2153G>A	p.R668H	uc004csf.1	P80
KNTC1	9735	121639218	Frame_Shift_Del	c.4064_4064delG	p.G1301fs	uc001ucv.1	P80
LAD1	3898	199622274	Missense	c.1073G>A	p.A280T	uc001gwm.1	P80
LRRK1	79705	99410672	Missense	c.4031C>G	p.L1238V	uc002bw.1	P80
LRRN4CL	221091	62212012	Missense	c.852C>G	p.P182R	uc001nun.1	P80
MYH6	4624	22935397	Missense	c.2432C>T	p.R789C	uc001wjv.2	P80
NINJ1	4814	94936257	Missense	c.135C>A	p.P22T	uc004atg.2	P80
NPR3	4883	32748081	Missense	c.660T>C	p.S148P	uc003jhv.1	P80
PLB1	151056	28705507	Missense	c.3769G>T	p.A1257S	uc002rbm.1	P80
PTPRZ1	5803	121403502	Missense	c.891T>A	p.F166I	uc003vjy.1	P80
RAPGEF5	9771	22297387	Splice_Site_Ins	c.e6_splice_site		uc003svg.1	P80
SENP6	26054	76463931	Missense	c.2885A>G	p.I756V	uc003pid.2	P80
SHANK1	50944	55867161	Missense	c.2619G>A	p.R867H	uc002psx.1	P80
SIM2	6493	37035984	Missense	c.1003A>C	p.N316T	uc002yvr.1	P80
SLC22A17	51310	22887326	Missense	c.778G>A	p.R241Q	uc001wj1.1	P80
TLE2	7089	2964816	Missense	c.847G>A	p.D243N	uc010dth.1	P80
TNPO2	30000	12678017	Missense	c.2399A>C	p.N646T	uc002mup.1	P80
UCK1	83549	133394153	Missense	c.696C>T	p.P201L	uc004cay.1	P80
ZMYND15	84225	4593457	Missense	c.1285C>A	p.H419N	uc002fyu.1	P80
ADCY1	107	45628857	Missense	c.1028A>G	p.E337G	uc003tne.2	P81
APOC2	344	50144278	Missense	c.341G>C	p.A80P	uc002pah.1	P81
ARID4B	51742	233411660	Missense	c.3695A>G	p.D1066G	uc001hwq.1	P81
ATP2B3	492	152483672	Missense	c.3385G>A	p.E1087K	uc004fht.1	P81

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C14orf183	196913	49620246	Missense	c.848T>C	p.L283S	uc001wxm.1	P81
C17orf82	388407	56844386	Missense	c.493G>T	p.G90W	uc002izh.1	P81
C22orf42	150297	30877000	Missense	c.513T>C	p.S158P	uc003amd.1	P81
CAMKK2	10645	120182506	Missense	c.919G>T	p.M265I	uc001tzu.1	P81
CD74	972	149772471	Missense	c.55A>G	p.D12G	uc003lsl.1	P81
CLDN1	9076	191513372	Missense	c.591C>T	p.A124V	uc003fsh.1	P81
EXOC3L	283849	65776586	Missense	c.1944G>T	p.G568V	uc002erx.1	P81
FAM116B	414918	49097454	Frame_Shift_Del	c.548_548delT	p.L102fs	uc003bkx.1	P81
HSD17B6	8630	55462229	Missense	c.628T>C	p.V173A	uc001smg.1	P81
IDH1	3417	208812085	Missense	c.1355G>A	p.G370D	uc002vcs.1	P81
KNDC1	85442	134877599	Missense	c.4661A>C	p.T1554P	uc001llz.1	P81
MCM6	4175	136325461	Missense	c.1974G>C	p.R633P	uc002tuw.1	P81
NALCN	259232	100827062	Missense	c.823A>G	p.T212A	uc001vox.1	P81
PLA2G4A	5321	185129859	Missense	c.476T>A	p.L91I	uc001gsc.1	P81
PLA2G4A	5321	185182666	Missense	c.1419T>G	p.L405W	uc001gsc.1	P81
RYR2	6262	236038954	Missense	c.14549T>C	p.L4810P	uc001hy1.1	P81
SCN7A	6332	167030735	Missense	c.800T>A	p.S225T	uc002udu.1	P81
SIRPA	140885	1843955	Missense	c.299C>A	p.T97K	uc002wft.1	P81
SLC22A7	10864	43374283	Missense	c.308C>T	p.P70L	uc003out.1	P81
SLT2	9353	20139695	Missense	c.1692A>C	p.L496F	uc003gpr.1	P81
SULT1A2	6799	28514733	Missense	c.371T>C	p.I77	uc002dqg.1	P81
TECTA	7007	120528887	Missense	c.4193G>C	p.C1398S	uc001pxr.1	P81
TEX15	56154	30823876	Missense	c.2200G>A	p.E734K	uc003xil.1	P81
TPRX1	284355	52997355	In_frame_Del	c.773_796delGAA	p.234_242PNPGPIP	uc002php.1	P81
ALKBH1	8846	77244084	Missense	c.26C>G	p.A6G	uc001xuc.1	P82
ATP1A4	480	158395908	Missense	c.1225A>C	p.N249T	uc001fve.2	P82
BCOR	54880	39818154	Frame_Shift_Del	c.1680_1681delCC	p.P463fs	uc004den.2	P82
BRSK2	9024	1389377	Missense	c.420T>C	p.L56P	uc001itm.2	P82
CAND1	55832	65961968	Missense	c.517C>A	p.T27K	uc001stn.2	P82
DNAH10	196385	122965416	Missense	c.10310G>C	p.A3429P	uc001uft.2	P82
DNAH9	1770	11588941	Missense	c.6282C>T	p.R2072C	uc002gne.1	P82
ENPEP	2028	111688855	Frame_Shift_Del	c.2417_2417delG	p.W692fs	uc003iab.2	P82
GRM5	2915	87940496	Missense	c.2203G>A	p.R668H	uc001pcq.1	P82
KIAA0430	9665	15610904	Missense	c.4124G>A	p.E1311K	uc002ddr.1	P82
KIAA0802	23255	8708540	Missense	c.234G>A	p.R31Q	uc002knr.2	P82
KIRREL3	84623	125800111	Nonsense	c.1997C>A	p.Y637*	uc001fea.1	P82
MAP1B	4131	71526245	Missense	c.1548A>G	p.Y436C	uc003kbw.2	P82
MEMO1	51072	31948527	Splice_Site_SNP	c.e8_splice_site	uc002rmx.1	P82	
MMP12	4321	102244005	Frame_Shift_Ins	c.674_675insA	p.T210fs	uc001phk.1	P82
NOTCH1	4851	138510470	Frame_Shift_Del	c.7541_7542delCT	p.P2514fs	uc004chz.1	P82
PPM1F	9647	20615657	Missense	c.868C>G	p.Q252E	uc002zvp.1	P82
PTH2	113091	54618366	Missense	c.145C>G	p.L15V	uc002prn.1	P82
SCAPER	49855	74808099	Missense	c.2091C>G	p.A685G	uc002bby.1	P82
SEC16B	89866	176196666	Missense	c.1688G>A	p.M274I	uc001glj.1	P82
SETBP1	26040	40785584	Missense	c.2577G>A	p.V707M	uc010dn1.i	P82
SMCR7	125170	18108688	Missense	c.1440T>C	p.L417P	uc002gst.1	P82
TSNAXIP1	55815	66412286	Missense	c.423C>A	p.T10K	uc002euj.1	P82
TTC7A	57217	47132436	Missense	c.2505A>G	p.M713V	uc010fb.1	P82
VARS	7407	31854805	Missense	c.4067C>G	p.A1215G	uc003nxe.1	P82
WWC1	23286	167804132	Missense	c.2555A>G	p.E830G	uc003lzu.1	P82
ZP4	57829	236115706	Missense	c.943C>T	p.L315F	uc001hym.1	P82
ATG9B	285973	150352416	Frame_Shift_Ins	c.103_104insG	p.G9fs	uc010lpv.1	P83
C11orf35	256329	545379	Missense	c.1762A>C	p.T567P	uc001lpx.1	P83
CNTROB	116840	7780825	Nonsense	c.1712C>T	p.Q265*	uc002gjp.1	P83
EVC	2121	5784069	Missense	c.585C>T	p.S134F	uc003gil.1	P83
FLNB	2317	58063032	Missense	c.1573C>T	p.R470W	uc010hne.1	P83
HYDIN	54768	69499816	Missense	c.8361G>T	p.V2745L	uc002ezr.1	P83
IFLTD1	160492	25564170	Missense	c.992G>A	p.R281H	uc001rgs.1	P83
IRF2	3660	185548886	Frame_Shift_Del	c.902_906delAAC	p.E234fs	uc003iwf.2	P83
MADCAM1	8174	452762	Missense	c.771A>C	p.Q254P	uc002los.1	P83
PANK4	55229	2436988	Missense	c.1256A>G	p.E416G	uc001ajm.1	P83
PDE2A	5138	71970570	Missense	c.2082C>T	p.R641W	uc001osm.1	P83
PRKG1	5592	53711936	Frame_Shift_Del	c.1680_1680delA	p.A521fs	uc001ijo.2	P83
PXDNL	137902	52547417	Missense	c.796A>G	p.Q232R	uc003xqu.2	P83
SAMD5	389432	147871912	Missense	c.157G>C	p.R52P	uc003qmc.1	P83
TMEM59	9528	54270424	Missense	c.1212A>G	p.H321R	uc001cwq.1	P83
TRPV4	59341	108705926	Missense	c.2594C>A	p.N833K	uc001tpj.1	P83
TTN	7273	179171917	Missense	c.49285A>G	p.E16354G	uc002umr.1	P83
TXLNB	167838	139633330	Nonsense	c.756G>T	p.E215*	uc010kha.1	P83
ANGPT2	285	6353944	Missense	c.1576T>C	p.I416T	uc003wj.2	P84
EVC2	132884	5675411	Missense	c.2309G>A	p.R752Q	uc003gij.1	P84
MICALCL	84953	12272963	In_frame_Del	c.1700_1702delCT	p.T471del	uc001mkg.1	P84
OR5AS1	219447	55555423	Missense	c.953G>A	p.R318H	uc001nif.1	P84
OR8J3	81168	55661275	Missense	c.496G>A	p.V166M	uc001nij.1	P84

Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
PGM5	5239	70189275	Missense	c.795T>A	p.F189Y	uc004agr.1	P84
PHLPP1	23239	58648424	Missense	c.395C>A	p.L73M	uc002lis.1	P84
PIWIL4	143689	93940400	Missense	c.219G>C	p.R23P	uc001pfa.1	P84
RECQL5	9400	71138513	Splice_Site_Ins	c.e12_splice_site		uc010dg1.1	P84
SF3B1	23451	197974954	Missense	c.2271G>T	p.K741N	uc002uuue.1	P84
SLC22A13	9390	38292790	Missense	c.1295G>A	p.V416M	uc003chz.2	P84
XPO1	7514	61572976	Missense	c.1840G>A	p.E571K	uc002sbi.1	P84
AGPAT9	84803	84744924	Missense	c.1503G>A	p.G429S	uc003how.1	P85
ATM	472	107677584	Splice_Site_SNP	c.e35_splice_site		uc001pkb.1	P85
CDHR3	222256	105440555	Missense	c.1144G>A	p.E356K	uc003vdl.2	P85
CHD9	80205	51899194	Missense	c.7045C>T	p.S2294F	uc002ehb.1	P85
CIDEB	27141	23845524	Missense	c.356G>C	p.E78Q	uc001won.1	P85
CXorf26	51260	75311728	Missense	c.378C>T	p.P59S	uc004ecl.1	P85
F8	2157	153744594	Missense	c.6703C>T	p.R2178C	uc004fmt.1	P85
MAN1C1	57134	25816933	Missense	c.388C>G	p.P20A	uc001bkm.2	P85
MDC1	9656	30781379	Missense	c.4000C>G	p.T1187S	uc003nrg.2	P85
MNT	4335	2237491	Missense	c.1455G>C	p.Q401H	uc002fur.1	P85
NEK10	152110	27301134	Missense	c.2251C>A	p.N659K	uc003cdt.1	P85
NLGN2	57555	7259157	Missense	c.1076C>T	p.R335W	uc002ggt.1	P85
PKHD1L1	93035	110477524	Missense	c.1008G>A	p.V302I	uc003yne.1	P85
SF3B1	23451	197975079	Missense	c.2146A>G	p.K700E	uc002uuue.1	P85
SLC25A42	284439	19079734	Missense	c.680A>T	p.I177F	uc002nlf.1	P85
TBC1D26	353149	15582353	Missense	c.564C>T	p.A105V	uc010cov.1	P85
ZIC2	7546	99432896	Nonsense	c.577C>T	p.Q193*	uc001won.1	P85
ZNF711	7552	84412580	Missense	c.2400G>A	p.S505N	uc004eeq.1	P85
ACTRT1	139741	127013502	Missense	c.557T>C	p.L122P	uc004eum.1	P86
ACVR2A	92	148401270	Missense	c.1669T>C	p.M500T	uc002twg.1	P86
C1orf113	79729	36558374	Missense	c.1052C>T	p.S154L	uc001cah.1	P86
C8orf76	84933	124322660	Missense	c.139C>G	p.C36W	uc003yqc.1	P86
CAPN6	827	110381154	Missense	c.1078C>G	p.Q304E	uc004epc.1	P86
DCN	1634	90082554	Nonsense	c.377G>T	p.E95*	uc001tbs.1	P86
DDX11	1663	31133692	Splice_Site_SNP	c.e8_splice_site		uc001rjt.1	P86
DLGAP1	9229	3869890	Missense	c.246C>T	p.P60L	uc002kmf.1	P86
ERC2	26059	56158107	Missense	c.1499G>A	p.R415S	uc003dhr.1	P86
FAM132A	388581	1168345	Missense	c.723T>C	p.C231R	uc001adl.1	P86
FAM53B	9679	126301801	Read-through	c.1792A>G	p.*423W	uc001lhv.1	P86
GUCY1A2	2977	106393739	Missense	c.643G>A	p.V85I	uc009yxn.1	P86
KIF4A	24137	69489226	Missense	c.1610G>A	p.E495K	uc004dyg.1	P86
LGALS3	3958	54674798	Missense	c.452T>C	p.Y101H	uc001xbr.1	P86
MFSD7	84179	666077	Missense	c.1206C>G	p.P373R	uc003gb.1	P86
NBEAL2	23218	47015888	Missense	c.3802A>G	p.Q1208R	uc003ccp.2	P86
NOS1	4842	116252978	Missense	c.965G>C	p.V94L	uc001twm.1	P86
PRIC285	85441	61663879	Missense	c.7411G>T	p.Q2173H	uc002yfm.2	P86
ProSAP1P1	9762	3093302	Missense	c.3218A>G	p.E607G	uc002wia.1	P86
RPS28	6234	8292862	Missense	c.144C>G	p.T38R	uc002mjn.1	P86
SAMHD1	25939	34981271	Missense	c.892G>A	p.M254I	uc002xgh.1	P86
SEMA4C	54910	96890742	Missense	c.2240C>G	p.A670G	uc002sxg.2	P86
SLCO2A1	6578	135148892	Missense	c.1466_1467CC>T	p.P398F	uc003eqa.2	P86
USP6NL	9712	11545726	Missense	c.1301A>G	p.R420G	uc001iks.1	P86
YIPF3	25844	43591402	Missense	c.479A>G	p.K108E	uc010jyr.1	P86
ZMYM3	9203	70377817	Missense	c.3992T>C	p.F1302S	uc004dzd.1	P86
BCOR	54880	39819146	Frame_Shift_Del	c.688_689delGG	p.V132fs	uc004den.2	P87
C11orf16	56673	8905208	Missense	c.538A>T	p.L138F	uc001mh2.2	P87
C19orf35	374872	2226747	Missense	c.1448T>G	p.C452G	uc002lvn.1	P87
CEP350	9857	178297999	Missense	c.5667G>A	p.E1762K	uc001gnt.1	P87
GPR128	84873	101856634	Missense	c.1901G>C	p.A549P	uc003duc.1	P87
GRIN3A	116443	103379932	Missense	c.3548A>C	p.I983L	uc004bbp.1	P87
IGSF10	285313	152647512	Missense	c.2947C>T	p.P983S	uc003zeb.1	P87
INPP5D	3635	233633454	Missense	c.175G>A	p.G8S	uc002vtv.1	P87
KCNC2	3747	73730870	Missense	c.1726G>T	p.L394F	uc001sxg.1	P87
NCKAP5	344148	133257568	Missense	c.3660G>A	p.A1096T	uc002ttip.1	P87
NOTCH1	4851	138510470	Frame_Shift_Del	c.7541_7542delCT	p.P2514fs	uc004chz.1	P87
NR4A1	3164	50738769	Missense	c.2728T>G	p.V578G	uc001rzq.1	P87
OR2G6	391211	246752085	Missense	c.515G>A	p.R172H	uc001ien.1	P87
PBX2	5089	32262573	Missense	c.1379T>G	p.S370A	uc003oav.1	P87
PLEKHA5	54477	19327644	Missense	c.1465G>A	p.G487R	uc001rea.1	P87
TDRD5	163589	177897973	Missense	c.2629G>C	p.A812P	uc001ngn.1	P87
CAMK4	814	110740514	Missense	c.461G>A	p.V121I	uc003kpf.1	P88
GPR39	2863	132891393	Missense	c.777G>A	p.S103N	uc002ttl.1	P88
INPP4A	3631	98528924	Missense	c.1113A>G	p.N337S	uc002sy.1	P88
MYO15A	51168	17993350	Missense	c.7390G>C	p.S2351T	uc010cpt.1	P88
NRAS	4893	115058052	Missense	c.436A>G	p.Q61R	uc009wgu.1	P88
PIK3C2A	5286	17114687	Missense	c.1832A>G	p.D589G	uc001mmq.2	P88
PLK1	5347	23599822	Missense	c.717G>T	p.V222L	uc002dlz.1	P88

Gene Name	Gene ID	Start_position	Variant_Classification	cdDNA_Change	Protein_Change	Annotation	Patient ID
SAMHD1	25939	34973148	Missense	c.1287T>G	p.I386S	uc002xgh.1	P88
SLC27A5	10998	63714890	Frame_Shift_Del	c.268_268delC	p.P82fs	uc002qtc.1	P88
SOX8	30812	973775	Missense	c.584C>T	p.R157C	uc002ckn.1	P88
STX16	8675	56684652	Nonsense	c.1612G>T	p.E293*	uc002xzi.1	P88
TSC2	7249	2061594	Missense	c.2028G>T	p.S641N	uc002con.1	P88
ZNF146	7705	41419850	Missense	c.2191A>G	p.Q223R	uc002odq.2	P88
ZNF668	79759	30980681	Missense	c.1426G>T	p.V357L	uc010caf.1	P88
GALK2	2585	47249819	Missense	c.106C>A	p.T3K	uc001xjx.1	P89
MYH7B	57644	33046900	Missense	c.3019A>G	p.E976G	uc002xbi.1	P89
NFKBIA	4792	34943526	Missense	c.186C>A	p.L26M	uc001wtf.2	P89
PASD1	139135	150583294	Missense	c.1221T>C	p.Y297H	uc004fev.2	P89
PHKA2	5256	18825279	Missense	c.3635C>G	p.R1069G	uc004cyv.2	P89
SEMA4G	57715	102733150	Missense	c.2188C>A	p.L602I	uc001krw.1	P89
TCF3	6929	1583078	Missense	c.287G>C	p.S86T	uc002ltp.1	P89
TJP2	9414	71039274	Missense	c.1971C>G	p.R591G	uc004ahe.1	P89
VASH1	22846	76306148	Splice_Site_SNP	c.e2_splice_site		uc001xst.2	P89
DNAH1	25981	52379823	Missense	c.6543A>G	p.E2156G	uc003dds.1	P90
DNHD1	144132	6545248	Missense	c.6207G>C	p.R2032P	uc001mdw.2	P90
HACE1	57531	105305028	Nonsense	c.2501C>T	p.Q742*	uc003pqu.1	P90
HIST1H1D	3007	26342680	Missense	c.516A>G	p.K154R	uc003nhd.1	P90
ICA1L	130026	203361882	Missense	c.1323G>T	p.G387W	uc002uzh.1	P90
LGSN	51557	64053489	Missense	c.326G>A	p.V98M	uc003peh.1	P90
NOC2L	26155	881356	Nonsense	c.648C>T	p.Q197*	uc009vjq.1	P90
OGFR	11054	60915226	Missense	c.1849G>T	p.R605L	uc002ydj.1	P90
PGBD5	79605	228564713	Missense	c.350C>T	p.T117M	uc001htv.1	P90
ROBO1	6091	79070687	Missense	c.253G>A	p.A85T	uc003dqe.1	P90
SEMA3E	9723	82835175	Missense	c.2457C>T	p.T664M	uc003uhy.1	P90
TP53	7157	7518933	Missense	c.835A>G	p.H214R	uc002gim.2	P90
XRCC5	7520	216700595	Missense	c.923T>C	p.L297S	uc002fy.1	P90
ZNF142	7701	219217088	Nonsense	c.2831G>T	p.E799*	uc002vin.1	P90
ZNF579	163033	60781946	Missense	c.925T>G	p.V291G	uc002qlh.1	P90
ACSM2A	123876	20390445	Missense	c.1066C>A	p.P276H	uc010bw.1	P91
AFTPH	54812	64633697	Missense	c.1617A>G	p.K529E	uc002sdc.1	P91
C16orf57	79650	56611602	Missense	c.833A>C	p.Q250H	uc002emz.1	P91
C8orf47	203111	99170605	Missense	c.332T>A	p.L62I	uc003yi.1	P91
CELF3	11189	149946729	Missense	c.1444C>A	p.A217D	uc001eys.1	P91
DNHD1	144132	6536735	Missense	c.3513C>T	p.A1134V	uc001mdw.2	P91
F2R	2149	76064393	Missense	c.852T>C	p.I196T	uc003ken.2	P91
FAM50A	9130	153331803	Missense	c.1028T>A	p.I318N	uc004flk.1	P91
FNDC3B	64778	173495877	Missense	c.937T>A	p.L294M	uc010hwt.1	P91
GDF2	2658	48033667	Missense	c.1370G>A	p.V403I	uc001jfa.1	P91
GOLGA4	2803	37344179	Missense	c.6168C>G	p.A1955G	uc003cgw.1	P91
HCK	3055	30131240	Nonsense	c.602G>A	p.W144*	uc002wxh.1	P91
KIAA0467	23334	43671006	Missense	c.3316C>T	p.R952W	uc001cjk.1	P91
KIAA0947	23379	5516221	Frame_Shift_Del	c.3996_3999delTC	p.T1258fs	uc003jdm.2	P91
KRT17	3872	37033980	Missense	c.356G>A	p.R103H	uc002hxh.1	P91
MAGEC1	9947	140821627	Missense	c.1057G>C	p.Q257H	uc004fbt.1	P91
MLL	4297	117880825	Missense	c.9031A>T	p.D3003V	uc001ptb.1	P91
NIN	51199	50302815	Missense	c.1786G>C	p.R532T	uc001wyi.1	P91
NPC1	4864	19390537	Missense	c.1157G>C	p.E332Q	uc002kum.2	P91
OLR1	4973	10204214	Missense	c.793T>G	p.L227V	uc001qxo.1	P91
PDE1C	5137	31759666	Missense	c.2761A>C	p.K723Q	uc003tco.1	P91
POLRMT	5442	575894	Missense	c.1021A>T	p.Q322L	uc002lpf.1	P91
RBMX	27316	135785210	Splice_Site_SNP	c.e7_splice_site		uc004fae.1	P91
RNF150	57484	142008975	Missense	c.1861G>A	p.E403K	uc003iio.1	P91
SF3B1	23451	197975079	Missense	c.2146A>G	p.K700E	uc002uu.1	P91
SLC46A1	113235	23755946	Missense	c.992G>C	p.W299S	uc002hbf.1	P91
SYT15	83849	46382034	Missense	c.1361G>T	p.S403I	uc001jea.1	P91
TP53	7157	7518931	Missense_Mutation	c.643A>C	p.S215R	NM_000546	P91
TP53	7157	7513653	Read-through	c.1375G>T	p.*394L	uc002gim.2	P91
TRO	7216	54972506	Missense	c.2731C>T	p.T875M	uc004dtq.1	P91
VDAC2	7417	76650736	Splice_Site_SNP	c.e8_splice_site		uc001jxa.1	P91

**Table 2** Analysis of mutation rate in CLL in relation to clinical characteristics.

		Silent mutation rate		Non-silent mutation rate		Total mutation rate		
		N	Median, range	p-value*	Median, range	p-value*	Median, range	p-value*
Clinical Characteristics	Rai at sample			0.41		0.27		0.28
	0-1	72	0.19 (0.0, 1.09)		0.69 (0.08, 2.70)		0.88 (0.11, 3.79)	
	2-4	19	0.16 (0.04, 0.38)		0.57 (0.21, 1.25)		0.75 (0.29, 1.60)	
	Treatment status at sample			0.006		0.14		0.033
	Chemotherapy naïve	61	0.17 (0.0, 0.49)		0.66 (0.08, 1.44)		0.77 (0.11, 1.73)	
	Prior treatment	30	0.21 (0.07, 1.09)		0.70 (0.21, 2.70)		0.99 (0.29, 3.79)	
	Prior exposure to nucleoside analogue			0.005		0.088		0.019
	No	64	0.17 (0, 0.49)		0.64 (0.08, 1.44)		0.77 (0.11, 1.73)	
	Yes	27	0.22 (0.07, 1.09)		0.73 (0.21, 2.70)		1.00 (0.29, 3.79)	
	IGHV mutation status			0.28		0.5		0.32
	Unmutated	40	0.19 (0.04, 0.92)		0.69 (0.08, 2.14)		0.92 (0.11, 3.06)	
	mutated	38	0.17 (0, 1.09)		0.68 (0.11, 2.70)		0.82 (0.18, 3.79)	
FISH Cytogenetics	ZAP-70			0.64		0.99		0.86
	Negative	44	0.18 (0.04, 1.09)		0.69 (0.11, 2.70)		0.87 (0.18, 3.79)	
	Positive	38	0.16 (0, 0.92)		0.68 (0.08, 2.14)		0.88 (0.11, 3.06)	
	13q heterozygous deletion			0.70		0.66		0.59
	No	38	0.18 (0, 1.09)		0.63 (0.08, 2.70)		0.84 (0.11, 3.79)	
	Yes	53	0.17 (0.0, 0.92)		0.69 (0.11, 2.14)		0.87 (0.18, 3.06)	
	13q homozygous deletion			0.48		0.24		0.23
	No	79	0.18 (0, 1.09)		0.67 (0.08, 2.70)		0.81 (0.11, 3.79)	
	Yes	12	0.20 (0.10, 0.38)		0.77 (0.52, 1.07)		0.90 (0.71, 1.36)	
	Trisomy 12			0.98		0.66		0.84
	No	78	0.19 (0, 1.09)		0.67 (0.08, 2.70)		0.86 (0.11, 3.79)	
	Yes	13	0.17 (0.07, 0.49)		0.69 (0.35, 1.25)		0.77 (0.54, 1.68)	
Frequent Mutations	11q deletion			0.85		0.85		0.96
	No	69	0.18 (0.04, 1.09)		0.66 (0.14, 2.70)		0.86 (0.18, 3.79)	
	Yes	22	0.19 (0, 0.46)		0.69 (0.08, 1.25)		0.93 (0.11, 1.60)	
	17p deletion			0.035		0.12		0.07
	No	74	0.17 (0, 1.09)		0.67 (0.08, 2.70)		0.84 (0.11, 3.79)	
	Yes	17	0.21 (0.08, 0.92)		0.77 (0.49, 2.14)		1.11 (0.61, 3.06)	
	p53			0.41		0.14		0.17
	Unmutated	77	0.17 (0, 1.09)		0.66 (0.08, 2.70)		0.81 (0.11, 3.79)	
	Mutated	14	0.20 (0.04, 0.92)		0.78 (0.14, 2.14)		1.09 (0.18, 3.06)	
	SF3B1			0.69		0.57		0.61
	Unmutated	77	0.18 (0.04, 0.92)		0.68 (0.08, 2.14)		0.86 (0.11, 3.06)	
	Mutated	14	0.20 (0, 1.09)		0.63 (0.40, 2.70)		0.83 (0.50, 3.79)	
Sequencing Source Material	ATM			0.80		0.53		0.78
	Unmutated	83	0.18 (0, 1.09)		0.69 (0.08, 2.70)		0.86 (0.11, 3.79)	
	Mutated	8	0.19 (0.07, 0.46)		0.58 (0.42, 1.25)		0.76 (0.59, 1.60)	
	MYD88			0.61		0.84		0.70
	Unmutated	82	0.18 (0, 1.09)		0.68 (0.08, 2.70)		0.86 (0.11, 3.79)	
	Mutated	9	0.19 (0.04, 0.47)		0.59 (0.38, 1.26)		0.74 (0.47, 1.73)	
	NOTCH1			0.41		0.94		0.81
	Unmutated	87	0.19 (0, 1.09)		0.67 (0.08, 2.70)		0.86 (0.11, 3.79)	
	Mutated	4	0.14 (0.07, 0.27)		0.65 (0.53, 0.92)		0.74 (0.70, 1.19)	
	DDX3X			0.17		0.30		0.18
	Unmutated	88	0.19 (0.04, 1.09)		0.69 (0.08, 2.70)		0.87 (0.11, 3.79)	
	Mutated	3	0.12 (0, 0.19)		0.57 (0.55, 0.58)		0.70 (0.55, 0.76)	
Sequencing Source Material	MAPK1**							
	Unmutated	89	0.18 (0, 1.09)	NA	0.67 (0.08, 2.70)	NA	0.86 (0.11, 3.79)	NA
	Mutated	2	(0.27, 0.36)		(0.34, 0.82)		(0.61, 1.18)	
	FBXW3**			0.74		0.37		0.36
	Unmutated	88	0.18 (0, 1.09)		0.67 (0.08, 2.70)		0.86 (0.11, 3.79)	
	Mutated	3	0.25 (0.08, 0.29)		0.74 (0.69, 0.90)		0.99 (0.77, 1.19)	
	ZMYM3			0.12		0.83		0.94
	Unmutated	87	0.19 (0, 1.09)		0.67 (0.11, 2.70)		0.86 (0.18, 3.79)	
	Mutated	4	0.09 (0.04, 0.25)		0.79 (0.08, 0.87)		0.94 (0.11, 0.99)	
	Whole genome amplified DNA (for exomes)			0.33		0.31		0.28
	No	40	0.20 (0.04, 1.09)		0.70 (0.14, 2.70)		0.90 (0.18, 3.79)	
	Yes	51	0.16 (0, 0.92)		0.67 (0.08, 2.14)		0.77 (0.11, 3.06)	
Sequencing Source Material	Source of germline DNA			0.01		0.006		0.006
	Buccal epithelia	80	0.18 (0, 1.09)		0.69 (0.29, 2.70)		0.87 (0.33, 3.79)	
	Skin fibroblasts	7	0.29 (0.08, 0.46)		0.67 (0.21, 1.12)		1.13 (0.29, 1.41)	
	Granulocytes	4	0.05 (0.04, 0.17)		0.13 (0.08, 0.42)		0.18 (0.11, 0.59)	

\*Testing excludes unknown category. \*\*One patient had two mutations of the same gene.

**Table 3.** Calculation of background rate of non-synonymous mutation in CLL.

<i>Category</i>	<i>Rate</i>
CpG transition	1.91 E-06
Other C:G transition	2.24 E-07
A:T transition	2.05 E-07
Any transversion	2.90 E-07
Indel + null	1.33 E-07
<b>Total</b>	<b>7.25 E-07</b>

**Table 4:** Summary of mutations that have been previously identified in the COSMIC database (v76) in the significantly mutated genes.

Gene	Total number samples examined	Total number mutations	# cases per mutation	AA change	Breast	Endometrial	Ovary	Pancreas	GIColon	Melanoma	Lung	Lymphoid neoplasms	DLBCL	Burkitts lymphoma	MALT lymphoma	ALL other*
<i>SF3B1</i>	93	6	1	p.Q534P	■											
			1	p.L1211L		■										
			1	p.R568H			■									
			1	p.Q699H				■								
			1	p.K700E					■							
			1	p.P718L						■						
<i>MYD88</i>	445	12	2	p.V217F												
			1	p.W218R												
			2	p.I220T												
			11	p.S219C												
			2	p.S222R												
			3	p.M232T												
			5	p.S243N												
			64	p.L265P												
			1	p.V52M												
			1	p.S149G												
			1	p.S149I												
			1	p.T294P												
<i>FBXW7</i>	5385	84	1	p.A315T												
			1	p.C386W												
			1	p.D130fs*41												
			1	p.D440N												
			1	p.D480Y												
			1	p.D520N												
			1	p.D527G												
			1	p.E110*												
			1	p.E117del												
<i>TP53BP1</i>	5385	84	1	p.E117del												
			1	p.E121Y												
			1	p.E693K												
			1	p.F549fs*6												
			1	p.G397D												
			1	p.G423V												
			2	p.G423V												
			1	p.G423V												
			1	p.G579_Q581>E												
<i>PTEN</i>	5385	84	1	p.H379R												
			1	p.H420Y												
			1	p.H460R												
			1	p.H470P												
			1	p.H540Y												
			1	p.I435fs*9												
			1	p.I563T												
			1	p.K11R												
			1	p.K164*												
<i>SMAD4</i>	5385	84	1	p.K371fs*7												
			1	p.K444fs*32												
			1	p.L288fs*45												
			1	p.L403fs*34												
			1	p.L594F												
			1	p.L651*												
			1	p.M467fs*5												
			1	p.P298R												
			2	p.P298S												
<i>AKT1</i>	5385	84	1	p.Q156E												
			1	p.Q220*												
			1	p.Q264R												
			1	p.Q303*												
			1	p.Q98*												
			1	p.R13*												
			3	p.R224*												
			6	p.R278*												
			1	p.R312S												
<i>NRAS</i>	5385	84	1	p.R367*												
			1	p.R367*												
			4	p.R393*												
			1	p.R393*												
			1	p.R441W												
			28	p.R465C												
			10	p.R465C												
			6	p.R465C												



Gene	Total number samples examined	Total number mutations	# cases per mutation	AA change	Breast	Endometrial	Ovary	Pancreas	G/colon	Melanoma	Lung	Lymphoid neoplasms	DLBCL	Burkitts lymphoma	MALT lymphoma	ALL other*
			2	p.D1682Y												
			9	p.D1853N	■											
			1	p.D1853V												
			1	p.D2708N												
			1	p.D2725G												
			2	p.D2725V												
			1	p.E1612_Q1620>*												
			1	p.E1991D												
			1	p.E2052*												
			1	p.E2164K												
			1	p.E2423G												
			1	p.E2423K												
			1	p.E26fs*7												
			2	p.E522fs*43												
			1	p.E770*												
			2	p.E848Q												
			1	p.F1209fs*19												
			1	p.F1463L												
			1	p.F1463S												
			1	p.F168_V170>L												
			1	p.F1683fs*7												
			1	p.F2732L												
			1	p.F2799fs*4												
			1	p.F570S												
			3	p.F858L												
			1	p.G138R												
			1	p.G2023R												
			1	p.G2063E												
			2	p.G2695A												
			2	p.G2867E												
			1	p.G2925D												
			1	p.G2925V												
			1	p.G3051V												
			1	p.G558*												
			1	p.H1380Y												
			1	p.H2872Q												
			1	p.H996Q												
			1	p.I1237fs*2												
			2	p.I1332fs*27												
			1	p.I1407S												
			1	p.I1407T												
			1	p.I1469M												
			2	p.I1681V												
			1	p.I2055fs*33												
			1	p.I2076S												
			1	p.I2356F												
			1	p.I2888T												
			1	p.I352T												
			1	p.K1454N												
			1	p.K1994E												
			1	p.K2213fs*22												
			1	p.K2237fs*11												
			1	p.K2418_R2419insK												
			1	p.K2717M												
			1	p.K2810del												
			1	p.K3018N												
			1	p.K902fs*18												
			1	p.L1322I												
			1	p.L1322P												
			1	p.L1472F												
			1	p.L1708fs*6												
			1	p.L1764fs*12												
			2	p.L1794L												
			1	p.L1910H												
			1	p.L1939V												
			1	p.L2004R												
			1	p.L2417P												
			1	p.L2427R												
			1	p.L2445P												
			1	p.L2450fs*11												
			1	p.L2722R												
			2	p.L2890V												
			1	p.L2945fs*7												
			1	p.L3017P												
			1	p.L895fs*4												
			1	p.M1040V												
			1	p.M1916I												
			1	p.M1L												
			1	p.M2616I												
			1	p.M2805fs*1												
			1	p.M855fs*24												
			1	p.N1739T												
			1	p.N1801Y												
			1	p.N750K												
			1	p.P1054R												

Gene	Total number samples examined	Total number mutations	# cases per mutation	AA change	Breast	Endometrial	Ovary	Pancreas	GI/colon	Melanoma	Lung	Lymphoid neoplasms	DLBCL	Burkitts lymphoma	MALT lymphoma	ALL	other*
			1	p.P1829fs*5													
			1	p.P2699R													
			1	p.P2842R													
			4	p.P604S													
			1	p.Q1128R													
			1	p.Q1361*													
			1	p.Q162*													
			1	p.Q163*													
			1	p.Q2414*													
			1	p.Q2442P													
			2	p.Q2442P													
			1	p.Q2593*													
			1	p.Q466*													
			1	p.Q747H													
			1	p.R1086L													
			1	p.R1304fs*43													
			1	p.R2263S													
			1	p.R2273fs*37													
			1	p.R23Q													
			1	p.R2400fs*6													
			1	p.R2443*													
			1	p.R2443Q													
			2	p.R2443Q													
			1	p.R2453P													
			1	p.R2486G													
			1	p.R2713K													
			1	p.R2832C													
			1	p.R2871_H2872>S													
			1	p.R2912K													
			4	p.R3008C													
			4	p.R3008H													
			3	p.R3047*													
			2	p.R337C													
			1	p.R337H													
			2	p.R337S													
			1	p.R717W													
			1	p.S1179F													
			1	p.S151fs*2													
			1	p.S1770*													
			1	p.S1905L													
			1	p.S207C													
			1	p.S2375I													
			1	p.S2394L													
			1	p.S2408L													
			1	p.S2546_I2548del													
			1	p.S2859F													
			1	p.S707fs*29													
			2	p.S707P													
			1	p.S853*													
			1	p.S978P													
			1	p.T1735fs*11													
			1	p.T1743I													
			1	p.T1953R													
			1	p.T2396S													
			1	p.T2438K													
			1	p.T2611fs*10													
			2	p.T2666A													
			1	p.T2911del													
			2	p.T2947S													
			1	p.T935T													
			1	p.V1292_Q1331del													
			2	p.V1941L													
			1	p.V2424G													
			1	p.V245A													
			2	p.V410A													
			1	p.W1221*													
			1	p.W2845*													
			1	p.W308*													
			1	p.W393*													
			1	p.W57*													
			1	p.Y1392fs*7													
			1	p.Y1475C													
			1	p.Y1961C													
			1	p.Y2019S													
			1	p.Y2627fs*29													
			1	p.Y2817*													
			1	p.Y2954C													
			1	p.Y332C													



Gene	Total number samples examined	Total number mutations	# cases per mutation	AA change	Breast	Endometrial	Ovary	Pancreas	GI/colon	Melanoma	Lung	Lymphoid neoplasms	DLBCL	Burkitts lymphoma	MALT lymphoma	ALL	other*
			1	p.F1607_K1608insNPNVVLFK													
			1	p.F1607_K1608insVRVTHTK													
			1	p.F1607_R1609>LG													
			1	p.F1607>FVA													
			2	p.F1607>LD													
			1	p.F1607>LDP													
			1	p.F1607>LGM													
			1	p.F1607>LGT													
			1	p.F1607>LNPLS													
			1	p.F1607>LPHPHP													
			2	p.F1607>LPRNED													
			1	p.F1607>LRFL													
			1	p.F1607>LSMPP													
			1	p.F1607>WNS													
			1	p.F1618_P1619del													
			1	p.F1618_P1619insPFP													
			1	p.F1618_Y1620>WSP													
			1	p.F1618>FKN													
			3	p.F1618del													
			1	p.F1694S													
			1	p.F1737_M1738ins13													
			1	p.F2267fs*87													
			1	p.F2482fs*5													
			1	p.F2510fs*1													
			1	p.G1137V													
			1	p.G1216D													
			1	p.G135W													
			1	p.G1559V													
			1	p.G1647S													
			1	p.G1657S													
			1	p.G1660D													
			1	p.G1705*													
			2	p.G2153R													
			1	p.G2153S													
			1	p.G2246R													
			1	p.G2263fs*6													
			1	p.G2334fs*21													
			1	p.G2421fs*3													
			3	p.H1592_F1593>QT													
			1	p.H1592_F1593insY													
			1	p.H1592Y													
			1	p.H1602_T1603insQ													
			1	p.H1602P													
			1	p.H1612_G1613>QIVVFKRDA													
			1	HG													
			1	p.H1612_P1619>RGT													
			1	p.H2276fs*79													
			1	p.H2419fs*16													
			1	p.H2429fs*8													
			1	p.H2508Y													
			1	p.II617_R1623>G													
			7	p.II617N													
			1	p.II632V													
			1	p.II633I													
			1	p.II676_V1677>I													
			1	p.II676_V1677>MF													
			4	p.II676>TAFL													
			2	p.II681N													
			2	p.II681S													
			1	p.II719T													
			1	p.I2457fs*21													
			1	p.K1608_R1609insPAK													
			1	p.K1608>GPPLQ													
			1	p.K1608N													
			2	p.K1783_R1784ins31													
			31	p.L122fs*3													
			2	p.L1575P													
			1	p.L1575Q													
			35	p.L1586>PPEAV													
			1	p.L1586P													
			1	p.L1594_R1595ins12													
			1	p.L1594_R1595insA													
			1	p.L1594>NPM													
			34	p.L1594P													
			1	p.L1597_S1598insG													
			3	p.L1597H													
			1	p.L1601_H1602insA													
			1	p.L1601_H1602insL													
			40	p.L1601P													
			5	p.L1601Q													
			29	p.L1679P													
			5	p.L1679Q													
			1	p.L1707_A1708ins14													
			4	p.L1710P													
			1	p.L2327fs*5													
			1	p.L2336fs*19													
			1	p.L2336fs*20													
			1	p.L2343fs*12													

Gene	Total number samples examined	Total number mutations	# cases per mutation	AA change	Breast	Endometrial	Ovary	Pancreas	GI/colon	Melanoma	Lung	Lymphoid neoplasms	DLBCL	Burkitts lymphoma	MALT lymphoma	ALL	other*		
				p.L2391_Q2392>PFPF* p.L2430_G2431>CLVSR p.L2435fs*1 p.L2435fs*2 p.L2447fs*33 p.L2458V p.L2465L p.L2469fs*10 p.L2469fs*11 p.L2473fs*1 p.L2473fs*7 p.L2511L p.M1581_P1582del p.M1581_P1582insLMHLAF p.M1581_P1582insPRYEL p.M1581del p.M1616_F1618>L p.M1738_Y1739ins35 p.M2057fs*211 p.M2347fs*16 p.M2347fs*9 p.N1900I p.N2296_F2297insWV p.N2390fs*33 p.N2402_I2403>GPSLNN p.N2402fs*21 p.P1582_E1584>Q p.P1583_E1584insP p.P1583_L1586>IEA p.P1583del p.P2272S p.P2333fs*22 p.P2411fs*12 p.P2412del p.P2412P p.P2413S p.P2413T p.P2414L p.P2416del p.P2418L p.P2439fs*40 p.P2439fs*41 p.P2439L p.P2459fs*21 p.P2459fs*61 p.P2463fs*15 p.P2475fs*1 p.P2475fs*3 p.P2475fs*34 p.P2475fs*5 p.P2476fs p.P2476fs*2 p.P2494fs*13 p.P2494fs*3 p.P2506fs*6 p.P2506P p.P2509fs*8 p.P2513fs*3 p.P2513L p.P2515fs p.P2515fs*4 p.P2518* p.P2518fs*6 p.Q1050L p.Q1585>PVELMPPE p.Q1585del p.Q1615_F1618>LCR p.Q1615_M1616>L p.Q1615K p.Q1685_C1686insLEGQR p.Q2316* p.Q2344fs*11 p.Q2392* p.Q2394* p.Q2395* p.Q2396* p.Q2399* p.Q2404* p.Q2406* p.Q2407*															

Gene	Total number samples examined	Total number mutations	# cases per mutation	AA change	Breast Endometri- al Ovary Pancreas GI/colon Melanoma Lung Lymphoid neoplasms DLBCL Burkitts lymphoma MALT lymphoma ALL other*
			1	p.Q2410*	
			2	p.Q2417*	
			4	p.Q2441*	
			1	p.Q2445*	
			1	p.Q2445fs*65	
			9	p.Q2460*	
			2	p.Q2460fs*18	
			2	p.Q2502*	
			3	p.Q2504*	
			1	p.Q2504fs	
			1	p.Q2504fs*5	
			2	p.Q2520*	
			3	p.R1587P	
			3	p.R1595_E1596ins12	
			1	p.R1595_L1597>L	
			2	p.R1595>PRLPHNSSFHFLR	
			1	p.R1595>PRLPHNSSHFL	
			1	p.R1599>QS	
			20	p.R1599P	
			1	p.R1609_A1611>T	
			1	p.R1609_D1610ins12	
			1	p.R1609S	
			1	p.R1628H	
			1	p.R1628Q	
			1	p.R1634L	
			1	p.R1663L	
			1	p.R2160H	
			1	p.R2273fs*78	
			1	p.R2328W	
			1	p.S1598I	
			3	p.S1675_I1676insG	
			1	p.S1675P	
			1	p.S1709S	
			1	p.S2290R	
			1	p.S2291S	
			2	p.S2330fs*25	
			1	p.S2330fs*7	
			1	p.S2337fs*18	
			1	p.S2342fs*1	
			1	p.S2342fs*13	
			1	p.S2342fs*7	
			2	p.S2408N	
			1	p.S2423fs*1	
			1	p.S2424*	
			2	p.S2427fs*4	
			1	p.S2433fs*5	
			1	p.S2436fs*2	
			1	p.S2440fs*1	
			4	p.S2440fs*4	
			1	p.S2440G	
			1	p.S2450fs*28	
			1	p.S2468*	
			1	p.S2468fs*1	
			1	p.S2468fs*10	
			2	p.S2468fs*11	
			1	p.S2468fs*15	
			2	p.S2487*	
			1	p.S2487fs*7	
			1	p.S2492fs*67	
			3	p.S2493*	
			1	p.S2493>S*	
			1	p.S2493>SP*	
			1	p.S2493s*100	
			1	p.S2493fs*3	
			1	p.S2514F	
			1	p.S2514fs*4	
			4	p.S2524*	
			1	p.S2528fs*80	
			1	p.S356del	
			1	p.T1574_V1576del	
			1	p.T1603_N1604ins17	
			1	p.T1997M	
			1	p.T2467fs*11	
			1	p.T2467fs*12	
			1	p.T2467M	
			1	p.T2484A	
			2	p.T2484M	
			1	p.T2512fs*1	
			1	p.T445T	
			1	p.T971I	
			1	p.V1576_V1578del	
			1	p.V1577_V1578>FRP	
			1	p.V1577A	
			3	p.V1577E	
			1	p.V1578_V1579insA	
			1	p.V1578_V1579insGV	
			1	p.V1578A	
			1	p.V1579A	
			20	p.V1579del	

Gene	Total number samples examined	Total number mutations	# cases per mutation	AA change	Breast Endomet rial Ovary Pancreas GI/colon Melanom Lung Lymphoid neoplasm DLBCL Burkitts lymphom MALT lymphom ALL other*
ZMYM3	122	1	1	p.C883*	
			2	p.V1579E	
			1	p.V1579G	
			1	p.V1605_R1609>LKGCD	
			1	p.V1605_V1606del	
			1	p.V1605_V1606insN	
			1	p.V1605E	
			1	p.V1605G	
			1	p.V1606_F1607insLGR	
			1	p.V1606_F1607insLVY	
			1	p.V1606del	
			5	p.V1672I	
			1	p.V1677_Y1678insA	
			1	p.V1677>GIV	
			3	p.V1677D	
			1	p.V1677H	
			2	p.V1722_V1722ins?	
			1	p.V1722>ARWGSNLNIPYLI	
			1	p.V1722>PPGSL	
			1	p.V1722E	
			1	p.V1722G	
			4	p.V1722M	
			1	p.V1740_A1741ins14	
			1	p.V1740_A1741ins15	
			3	p.V2286I	
			1	p.V2331fs*23	
			1	p.V2422fs*2	
			1	p.V2422M	
			1	p.V2444A	
			1	p.V2444fs*27	
			2	p.V2444fs*3	
			1	p.V2444fs*34	
			11	p.V2444fs*35	
			2	p.V2444fs*36	
			6	p.V2444fs*37	
			1	p.V2444fs*39	
			1	p.V2444fs*69	
			1	p.V2444fs*73	
			1	p.V2454fs*25	
			1	p.V2474fs*4	
			1	p.V2474fs*5	
			1	p.V2537I	
			1	p.W2521*	
			1	p.Y1620_Y1621>PGG	
			1	p.Y1620N	
			1	p.Y1678>RAS	
			1	p.Y1717F	
			1	p.Y1739_V1740ins11	
			1	p.Y2491*	
			1	p.Y2491fs*1	

**Table 5.** Comparison of the clinical characteristics of the discovery (n=91) vs extension (n=101) samples.

	Discovery Cohort	Extension Cohort	p-value
<b>N</b>	91	101	
<b>Age at Diagnosis (years), median (range)</b>	54 (34, 78)	55 (30, 79)	0.5
Age ≥55 yrs.	40 (44)	52 (51)	0.31
<b>Sex</b>			
Female	35 (38)	51 (50)	0.11
Male	56 (62)	50 (50)	
<b>Time from Dx to 1st Therapy (months), median (range)</b>	30 (0.4, 154)	32 (1.3, 234)	0.7
<b># Patients initiating first therapy</b>	58 (64)	38 (38)	<0.001
<b>IGHV</b>			
Mutated	38 (42)	56 (55)	0.04
Unmutated	40 (44)	26 (26)	
Unknown	13 (14)	19 (19)	
<b>ZAP-70</b>			
Positive	38 (42)	33 (33)	0.17
Negative	44 (48)	49 (49)	
Unknown	9 (10)	19 (19)	
<b>FISH Cytogenetics</b>			
del (13q-) het	53 (58)	59 (58)	0.55
del (13q-) homo	12 (13)	0 (0)	<0.001
trisomy 12	13 (14)	15 (15)	0.84
del (11q)	22 (24)	11 (11)	0.03
del(17p)	15 (16)	14 (14)	0.84
Unknown	0 (0)	8 (8)	
<b>Somatic Mutations</b>			
<i>SF3B1-K700E</i>	7 (8)	3 (3)	0.2
<i>MYD88-P258L, L265P</i>	7 (8)	5 (5)	0.55
<i>NOTCH1-P2514fs</i>	4 (4)	8 (8)	0.38

**Table 6:** Additional mutations in the five core pathways. Genomic position and transcript annotation are provided with respect to genome build hg18 (<http://genome.ucsc.edu>).

Pathway	Gene Name	Gene ID	Start_position	Variant_Classification	cDNA_Change	Protein_Change	Annotation	Patient ID
DNA damage and Cell cycle control	ANAPC4	29945	24993979	Splice_Site_SNP	c.e4_splice_site		uc003gro.1	P19
	CDC14B	8555	98324609	Missense	c.1795C>G	p.T448R	uc004awj.1	P58
	PTTG1	9232	159781905	Missense	c.53C>A	p.T3N	uc003ly.1	P72
	ESPL1	9700	51949811	Missense	c.909G>A	p.S273N	uc001sck.2	P73
	HDAC4	9759	239701828	Missense	c.2426C>T	p.P645L	uc002vyk.2	P39
	E2F3	1871	20595009	Missense	c.1322T>C	p.I332T	uc003nda.2	P34
	CCNB3	85417	50107426	Missense	c.4170A>C	p.Q1291P	uc004dox.2	P69
	SMC1A	8243	53439984	Missense	c.2819C>A	p.T917N	uc004dsg.1	P57
	ERCC4	2072	13933620	Missense	c.1088A>G	p.K360R	uc002dce.2	P52
	BRCA1	672	38499191	Missense	c.2083G>A	p.S628N	uc002ict.1	P48
Inflammatory pathways	FANCA	2175	88385382	Missense	c.1331C>T	p.A430V	uc002fou.1	P31
	MSH4	4438	76086496	Missense	c.1218C>G	p.L393V	uc001dhd.1	P14
	CD14	929	139991681	Missense	c.1426T>C	p.S358P	uc003igl.1	P9
	TLR8	51311	12848204	Missense	c.1329G>T	p.R393I	uc004cvd.1	P52
	RIPK1	8737	3058352	Missense	c.2028A>G	p.K599R	uc010jni.1	P41
	MAP3K14	9020	40723695	Missense	c.309C>G	p.A67G	uc002iiv.1	P19
	MAPK8	5599	49303987	Missense	c.963G>A	p.E247K	uc009xnz.1	P1
	IRAK4	51135	42466478	Missense	c.1322A>G	p.K400E	uc001mu.2	P77
	TRAF3	7187	102408006	Splice_Site_SNP	c.e4_splice_site		uc001ymc.1	P15
	PPM1A	5494	59819255	Missense	c.396C>A	p.S100R	uc001xew.2	P27
RNA processing	NFKBIA	4792	34943526	Missense	c.186C>A	p.L26M	uc001wtf.2	P89
	IFNA8	3445	21399358	Missense	c.213C>G	p.F61L	uc003zpc.1	P39
	SPOP	8405	45051434	Missense	c.859G>A	p.D130N	uc002pb.1	P32
	PRPF8	10594	1524616	Missense	c.3283C>T	p.R1057W	uc002fte.1	P63
	RBM39	9584	33776456	Missense	c.796A>T	p.D151V	uc002xeb.1	P34
	U2AF2	11338	60864312	Missense	c.1486T>A	p.M144K	uc002qlu.1	P39
	CPSF2	53981	91678442	Missense	c.1080G>T	p.K281N	uc001yah.1	P2
	XPO1	7514	61572976	Missense	c.1840G>A	p.E571K	uc002sbi.1	P84

**Table 7.** Clinical characteristics of CLL patients harboring the 9 driver mutations.

**TP53**

Pt: Treatment status	Protein change	Mutation type	Cytogenetic abnormalities	ZAP70	IGHV
<b>Untreated</b>					
P74	L111R	Missense	del (17p)	No	Unmut
P62	R273C	Missense	None	No	Mut
P76	H193L	Missense	del(13q)	No	Mut
P49	N131del	In frame del	del(13q);del(17p)	Yes	Un
P90	H214R	Missense	del(17p)	N/A	N/A
<b>Treated</b>					
P3	R248Q	Missense	del (13q); del (17p)	Yes	Unmut
P9	I255F	Missense	Trisomy 12; del (13q); del (17p)	No	Unmut
P41	C238S	Missense	del (13q); del (17p)	No	Unmut
P42	D281N	Missense	Trisomy 12; del (13q); del (17p)	Yes	Mut
P91	S215R *394L	Missense Read through	del (13q)	Yes	Unmut
P72	G187_splice	Splice site	del (13q); del (11q); del (17p)	Yes	Unmut
P33	R273H	Missense	del(13q); del(17p)	Yes	Unmut
P39	C135Y	Missense	del(13q); del(17p)	Yes	Unmut
P65	R273H	Missense	Tri (12), del (13q); del (17p)	Yes	Unmut

**ATM**

Pt: Treatment status	Protein change	Mutation type	Cytogenetic abnormalities	ZAP70	IGHV
<b>Untreated</b>					
P8	L2135fs	Frame shift	None	N/A	Unmut
P17	Y1252F	Missense	del (13q)	No	Mut
P23	H2038R	Missense	Trisomy 12	Yes	N/A
<b>Treated</b>					
P5	Y2954C	Missense	del (13q); del (11q)	Yes	Mut
P73	Q2522H	Missense	Trisomy 12; del (13q)	Yes	N/A
	Y2817*	Stop	(13q); del (11q)		
P48	L546fs	Frame shift	Del (13q); del (11q)	Yes	Unmut
P85	C1726_splic_e	Splice site	Del (13q); del (11q)	Yes	Unmut
P61	K468fs	Frame shift	normal	No	N/A

**MYD88**

Pt: Treatment status	Protein change	Mutation type	Cytogenetic abnormalities	ZAP70	IGHV
<b>Untreated</b>					
P17	L265P	Missense	del (13q)	No	Mut
P18	M232T	Missense	del (13q)	No	Mut
P20	L265P	Missense	del (13q)	Yes	Mut

P25	L265P	Missense	Trisomy 12; del (13q)	No	Mut
P67	M232T	Missense	del (13q)	No	Mut
P31	L265P	Missense	del (13q)	Yes	Mut
<b>Treated</b>					
P5	L265P	Missense	del (13q); del (11q)	Yes	Mut
P46	P258L	Missense	del (13q); del (17p)	No	Mut
P66	L265P	Missense	del (13q)	No	Mut

### *SF3B1*

<i>Pt: Treatment status</i>	<i>Protein change</i>	<i>Mutation type</i>	<i>Cytogenetic abnormalities</i>	<i>ZAP70</i>	<i>IGHV</i>
<i>Untreated</i>					
P32	K700E	Missense	del (13q); del (11q)	No	Unmut
P8	G742D	Missense	None	N/A	Unmut
P37	K700E	Missense	del (11q)	Yes	Mut
P43	K700E	Missense	del (11q); del (17p)	Yes	Unmut
P51	G742D	Missense	del (11q)	N/A	N/A
P58	G740E	Missense	del (13q)	Yes	Unmut
P84	K741N	Missense	normal	No	Unmut
<b>Treated</b>					
P6	N626H	Missense	del (13q); del (11q)	No	Unmut
P40	Q903R	Missense	del (13q); del (11q)	Yes	Unmut
P60	R625L	Missense	del (13q); del (11q)	Yes	Unmut
P91	K700E	Missense	del (13q)	Yes	Unmut
P59	K700E	Missense	del (13q); del (17p)	Yes	Unmut
P61	K700E	Missense	normal	N/A	N/A
P85	K700E	Missense	Del (13q); del (11q)	Yes	Unmut

### *FBXW7*

<i>Pt: Treatment status</i>	<i>Protein change</i>	<i>Mutation type</i>	<i>Cytogenetic abnormalities</i>	<i>ZAP70</i>	<i>IGHV</i>
<i>Treated</i>					
P12	R505C	Missense	del (13q)	No	Mut
P35	G597E	Missense	del (11q)	Yes	Unmut
P42	F280L	Missense			
	R465H	Missense	del (13q); del (17p)	Yes	Mut

### *DDX3X*

<i>Pt: Treatment status</i>	<i>Protein change</i>	<i>Mutation type</i>	<i>Cytogenetic abnormalities</i>	<i>ZAP70</i>	<i>IGHV</i>
<i>Treated</i>					
P3	S24*	Nonsense	del (13q); del (17p)	Yes	Unmut
P6	K342_splice	Splice site	del (13q); del (11q)	No	Unmut
P37	S410fs	Frame shift	del (11q)	Yes	Mut

### *MAPK1*

<i>Pt: Treatment status</i>	<i>Protein</i>	<i>Mutation type</i>	<i>Cytogenetic abnormalities</i>	<i>ZAP70</i>	<i>IGHV</i>

	<i>change</i>					
<i>Treated</i>	Pt	Mutation	Type	Cytogenetic abnormalities	ZAP70	IGHV
	P29	Y316F	Missense	del (13q)	N/A	Mut
		D291G	Missense			
	P47	D162N	Missense	del (13q)	Yes	Unmut
<b><i>NOTCH1</i></b>						
<i>Pt: Treatment status</i>		<i>Protein change</i>	<i>Mutation type</i>	<i>Cytogenetic abnormalities</i>	<i>ZAP70</i>	<i>IGHV</i>
<i>Untreated</i>						
	P27	P2514fs	Frame shift	Tri (12)	No	N/A
	P82	P2514fs	Frame shift	Tri (12), del (13q); del (17p)	Yes	Unmut
<i>Treated</i>						
	P65	P2514fs	Frame shift	del (13q); del (17p)	Yes	Unmut
	P87	P2514fs	Frame shift	Tri (12), del (13q); del (11q)	yes	Unmut
<b><i>ZMYM3</i></b>						
<i>Pt: Treatment status</i>		<i>Protein change</i>	<i>Mutation type</i>	<i>Cytogenetic abnormalities</i>	<i>ZAP70</i>	<i>IGHV</i>
<i>Untreated</i>						
	P13	S1254T	Missense	del (13q)	N/A	Mut
	P86	F1302S	Missense	Normal	Yes	Unmut
	P38	S53fs	Frame shift	del(11q)	Yes	Unmut
<i>Treated</i>						
	P35	Q399*	Nonsense	del (13q)	Yes	Unmut

**Table 8.** Associations of driver mutations and (A) clinical characteristics and (B) FISH cytogenetics.

A.

Gene mutation	Gender			Age (years)			IGHV			ZAP70		
	Female	Male	p-value	<55	>=55	p-value	Unmutated	Mutated	p-value	Negative	Positive	p-value
<b>N</b>	35	56		51	40		40	38		44	38	
<b>p53</b>	5 (14)	9 (16)	0.99	8 (16)	6 (15)	0.99	10 (25)	3 (8)	0.07	5 (11)	8 (21)	0.36
<b>SF3B1</b>	7 (20)	7 (13)	0.38	8 (16)	6 (15)	0.99	9 (23)	2 (5)	0.048	5 (11)	8 (21)	0.36
<b>MYD88</b>	3 (9)	6 (11)	0.99	6 (12)	3 (8)	0.73	0 (0)	9 (24)	<0.001	6 (14)	3 (8)	0.49
<b>ATM</b>	2 (6)	6 (11)	0.71	5 (10)	3 (8)	0.99	3 (8)	2 (5)	0.99	2 (5)	6 (16)	0.14
<b>NOTCH1</b>	3 (9)	1 (2)	0.16	0 (0)	4 (10)	0.034	3 (8)	0 (0)	0.24	1 (2)	3 (8)	0.33
<b>ZMYM3</b>	2 (6)	2 (4)	0.64	4 (8)	0 (0)	0.13	4 (10)	0 (0)	0.12	0 (0)	4 (11)	0.042
<b>DDX3X</b>	0 (0)	3 (5)	0.28	1 (2)	2 (5)	0.58	2 (5)	1 (3)	0.99	1 (2)	2 (5)	0.59
<b>FBXW7</b>	2 (6)	1 (2)	0.56	1 (2)	2 (5)	0.58	1 (3)	2 (5)	0.61	1 (2)	2 (5)	0.59
<b>MAPK1</b>	1 (3)	1 (2)	0.99	2 (4)	0 (0)	0.5	1 (3)	1 (3)	0.99	0 (0)	1 (3)	0.46

B.

Gene mutation	del(13q) Het			del(13q) Homo			Trisomy 12			del(11q)			del(17p)		
	Negative	Positive	p-value	Negative	Positive	p-value	Negative	Positive	p-value	Negative	Positive	p-value	Negative	Positive	p-value
<b>N</b>	38	53		79	12		78	13		69	22		74	17	
<b>p53</b>	4 (11)	10 (19)	0.38	13 (16)	1 (8)	0.68	12 (15)	2 (15)	0.99	13 (19)	1 (5)	0.17	3 (4)	11 (65)	<0.001
<b>SF3B1</b>	7 (18)	7 (13)	0.56	13 (16)	1 (8)	0.68	14 (18)	0 (0)	0.21	6 (9)	8 (36)	0.004	13 (18)	1 (6)	0.45
<b>MYD88</b>	0 (0)	9 (17)	0.009	8 (10)	1 (8)	0.99	8 (10)	1 (8)	0.99	8 (12)	1 (5)	0.45	8 (11)	1 (6)	0.99
<b>ATM</b>	3 (8)	5 (9)	0.99	8 (10)	0 (0)	0.59	6 (8)	2 (15)	0.32	4 (6)	4 (18)	0.09	8 (11)	0 (0)	0.34
<b>NOTCH1</b>	1 (3)	3 (6)	0.64	4 (5)	0 (0)	0.99	1 (1)	3 (23)	0.009	3 (4)	1 (5)	0.99	2 (3)	2 (12)	0.16
<b>ZMYM3</b>	3 (8)	1 (2)	0.3	4 (5)	0 (0)	0.99	4 (5)	0 (0)	0.99	2 (3)	2 (9)	0.25	4 (5)	0 (0)	0.99
<b>DDX3X</b>	1 (3)	2 (4)	0.99	2 (3)	1 (8)	0.35	3 (4)	0 (0)	0.99	1 (1)	2 (9)	0.14	2 (3)	1 (6)	0.47
<b>FBXW7</b>	1 (3)	2 (4)	0.99	3 (4)	0 (0)	0.99	1 (1)	2 (15)	0.052	2 (3)	1 (5)	0.57	2 (3)	1 (6)	0.47
<b>MAPK1</b>	0 (0)	2 (4)	0.51	2 (3)	0 (0)	0.99	2 (3)	0 (0)	0.99	2 (3)	0 (0)	0.99	2 (3)	0 (0)	0.99

Note on multiple-hypothesis corrections:

q-value (1)=corrected for 9 hypotheses (the 9 possible genes being considered)

q-value (2)=corrected for 45 hypotheses (all combinations of genes x cytogenetic abnormalities)

**Table 9.** % Tumor cells harboring cytogenetic abnormalities.

Patient ID	del(13q) het	del(13q) homo	trisomy 12	del(11q)	del(17p)
P1	86	0	0	90	0
P2	0	0	0	0	0
P3	80	0	0	0	28
P4	0	46	0	0	0
P5	73	0	0	86	0
P6	40	10	0	15	0
P7	17	0	0	32	0
P8	0	0	0	0	0
P9	16	0	75	0	14
P10	10	0	0	0	0
P11	63	26	0	0	0
P12	16	0	35	0	8
P13	39	0	0	0	7
P14	88	0	0	0	0
P15	0	0	38	0	0
P16	0	89	0	0	0
P17	77	0	0	0	0
P18	30	0	0	0	0
P19	65	0	0	0	0
P20	61	0	0	0	0
P21	61	0	0	0	0
P22	10	0	0	0	6
P23	0	0	85	0	0
P24	0	90	0	0	0
P25	10	0	50	0	0
P26	0	27	0	0	0
P27	0	0	27	0	6
P28	83	0	0	0	0
P29	20	0	0	0	6
P30	20	0	0	0	0
P31	11	0	0	0	7
P32	24	0	0	89	0
P33	62	0	0	0	97
P34	20	0	0	33	0
P35	7	0	0	81	0
P36	30	0	0	43	0
P37	0	0	0	50	0
P38	0	0	0	72	0
P39	10	0	0	0	15
P40	16	0	0	27	0
P41	72	0	0	0	47
P42	72	0	18	0	86
P43	0	0	0	67	9
P44	0	0	0	0	46
P45	87	0	0	94	0
P46	26	51	0	0	11
P47	52	0	0	0	0
P48	96	0	0	91	0
P49	15	0	0	0	61
P50	0	0	0	0	0
P51	3	0	0	13	5
P52	6	91	0	0	0
P53	0	0	0	0	0
P54	36	7	0	0	0
P55	0	0	73	0	3

Patient ID	del(13q) het	del(13q) homo	trisomy 12	del(11q)	del(17p)
P56	0	0	0	0	0
P57	4	0	56	0	0
P58	24	0	0	0	0
P59	0	0	0	0	0
P60	93	0	0	34	0
P61	0	0	0	0	0
P62	0	0	0	0	0
P63	0	0	0	0	0
P64	0	82	0	0	9
P65	23	0	0	0	43
P66	24	0	0	0	0
P67	31	0	0	0	6
P68	61	0	0	0	0
P69	4	0	0	0	0
P70	0	61	0	0	0
P71	64	0	0	7	0
P72	97	0	0	19	46
P73	100	0	35	94	0
P74	0	0	0	0	45
P75	6	0	0	0	0
P76	6	40	0	0	0
P77	71	0	0	0	0
P78	25	0	0	29	29
P79	0	0	0	0	0
P80	81	0	0	0	0
P81	0	0	0	0	0
P82	9	0	32	0	12
P83	72	0	0	0	0
P84	5	0	0	0	0
P85	87	0	0	93	0
P86	0	0	0	0	0
P87	51	0	73	89	0
P88	0	0	76	0	0
P89	0	0	0	4	0
P90	0	0	0	0	47
P91	44	0	0	0	0

**Table 10:** Primers for the quantitative PCR of *BRD2* and *RIOK3* transcripts.

Target gene	Splicing status	Primers		
<i>BRD2</i>	Spliced	Applied Biosystems (Hs01121991_g1)		
	Unspliced	Forward	GCAAGATTTATACCATGTTCACCAACT	
		Reverse	CCCACCTACTAAATGAACACACAGA	
		Probe	CTCACCTTGTTGTAAATGT	
<i>RIOK3</i>	Spliced	Forward	CACAGCTTAGGCCTGAAGAAAA	
		Reverse	GCTGTCTTCATAAGGATGCACTTT	
		Probe	AAGGAAATGGAAACTTTG	
	Unspliced	Forward	CACAGCTTAGGCCTGAAGAAAA	
		Reverse	CCACTCAATGAAGTTGTACAATAAGG	
		Probe	CAATGGAGATAGCAAAGGTATT	

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